
NATIONAL NEWBORN SCREENING REPORT - 2000

Final Report: February, 2003



National Newborn Screening Report - 2000

(with Selected Totals)

A cooperative effort of the National Newborn Screening and Genetics Resource Center (NNSGRC) and the Association of Public Health Laboratories (APHL).

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Produced and Published by:

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Austin, Texas 78757
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February 2003

Single copies available at no charge from:

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Supported in part by Project #5U93MC00148-03 from the Maternal and Child Health Program (Title V, Social Security Act) Health Resources and Services Administration, United States Department of Health and Human Services.

ACKNOWLEDGMENTS

The editors acknowledge the State and Territorial Newborn Screening Coordinators, Laboratory Directors and the Association of Public Health Laboratories for their assistance in responding to the requests for data.

Appreciation is also extended to all members of the NNSGRC Newborn Screening Committee for their participation in the development of this report. Thanks also to the members of the newborn screening community and APHL for their continued interest and to those who assisted with assembly and review of the draft and final documents.

CITATION

National Newborn Screening and Genetics Resource Center, National Newborn Screening Report – 2000, NNSGRC, Austin, TX, February 2003

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Introduction

The National Newborn Screening and Genetics Resource Center

The National Newborn Screening and Genetics Resource Center (NNSGRC) is a funded project of the Health Resources and Services Administration (HRSA). The project is a cooperative agreement between the Maternal and Child Health Bureau, Genetic Services Branch and the University of Texas Health Science Center at San Antonio, Department of Pediatrics. **The mission of the NNSGRC is to: (1) provide a forum for interaction between consumers, health care professionals, researchers, organizations, and policy makers in refining and developing public health newborn screening and genetics programs, and (2) serve as a national resource center for information and education in the areas of newborn screening and genetics.**

The NNSGRC is established on the principle that all individuals are entitled to genetic literacy and genetic services in order to prevent or ameliorate the consequences of genetic disease. Accurate genetic information and competent genetic services should be provided to all individuals in a culturally competent, community based, family focused, prevention oriented, non-directive and confidential manner. To this end, states have a critical role to play in ensuring non-discriminatory effective genetic service and information delivery.

The NNSGRC serves as a focal point for national newborn screening and genetics activities, and provides related resources to benefit health professionals, the public health community, consumers and government officials. It also

serves to assist states in refining their newborn screening activities through technical assistance reviews and to enhance their capacity to incorporate new developments in genetics, health promotion and disease prevention into the public health system. In addition to annual national information reports on state and territorial newborn screening activities, the NNSGRC coordinates and facilitates national discussions of pertinent topics in the areas of newborn screening and genetics, and assists in developing and implementing related demonstration projects of national interest.

The NNSGRC is a component of the Genetics Division of the Department of Pediatrics, UTHSCSA.

NNSGRC Committees

The operational activities of NNSGRC draw on the expertise of two advisory committees. The Newborn Screening Committee and the Genetics Advisory Committee. Additionally there is a Newborn Screening Technical Review Team available to assist states in evaluating their newborn screening programs upon invitation of the State Health Department.

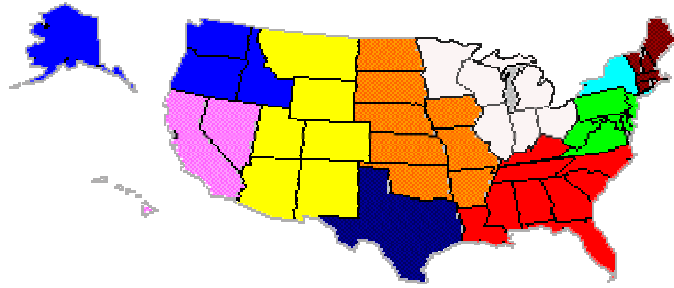
The NNSGRC committees are designed to study, plan, evaluate, report, recommend and implement projects and programs that are priorities of the NNSGRC.

Association of Public Health Laboratories

The Association of Public Health Laboratories (APHL), formerly the Association of State and Territorial Public Health Laboratory Directors (ASTPHLD), is a non-profit educational organization founded in 1951. APHL is dedicated to a healthier world through quality laboratory practice. The mission of APHL is to promote the role of public health laboratories in support of national and global objectives and to promote policies and programs which assure continuous improvement in the quality of laboratory practices. APHL works collaboratively with its members and others to reach common goals in pursuit of this mission. APHL maintains its national office in

Washington, D.C., and supports seven regional training offices of the National Laboratory Training Network (NLTN) in conjunction with the Centers for Disease Control and Prevention (CDC).

The prominence of newborn screening as a major public health laboratory assessment function has led to APHL's cooperation in ensuring the highest quality of laboratory data contained in this 2000 report.



Regional Networks for Genetic Services

(This diagram of previous CORN regions is for reference information only)

- | | | | |
|----------|-----------|------------|----------|
| ◆ GENES | ◇ GLaRGG | ◆ GPGSN | ◆ MARHGN |
| ◆ MSRGSN | ◆ NERGG | ◆ PacNoRGG | ◆ PSRGN |
| ◆ SERGG | ◆ TEXGENE | | |

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The NNSGRC Newborn Screening Committee (2000)

The NNSGRC's Newborn Screening Committee consists of one representative from each of the ten regional networks outlined on the map. The NNSGRC provides staff including, data coordination through the NNSGRC administrative assistant. Listed below are the committee representatives, their respective regions and the states and/or territory included in each region.

Newborn Screening Committee Members and Regions/Organizations Served

Randy Heidenreich, M.D. 520-626-5175	Mountain States Regional Genetic Services Network - MSRGSN Arizona, Colorado, Montana, New Mexico, Utah, and Wyoming
Gary Hoffman 608-262-4692	Great Lakes Regional Genetics Group - GLaRGG Illinois, Indiana, Michigan, Minnesota, Ohio and Wisconsin
Fred Lorey, Ph.D. 510-540-2941	Pacific Southwest Regional Genetics Group - PSRGN California, Hawaii, and Nevada
Fay Larson, R.N., M.S. 860-509-7499	New England Regional Genetics Group - NERGG Connecticut, Maine, Massachusetts, New Hampshire, Rhode Island, and Vermont
Charles Myers, M.S.W. 504-568-5070	Southeast Regional Genetics Group - SERGG Alabama, Georgia, Florida, Kentucky, Louisiana, Mississippi, North Carolina, South Carolina, and Tennessee
Kenneth A. Pass, Ph.D. 518-473-1993	Genetics Network of the Empire State - GENES New York, Puerto Rico, and Virgin Islands
Lou Bartoshesky 302-651-5916	Mid-Atlantic Regional Human Genetics Network - MARHGN Delaware, District of Columbia, Maryland, New Jersey, Pennsylvania, Virginia and West Virginia
Stuart Shapira, M.D., PH.D. 512-458-7430	Texas Genetics Network - TEXGENE Texas
Judi Tuerck, RN 503-494-7859	Pacific Northwest Regional Genetics Group - PacNoRGG Alaska, Idaho, Oregon, and Washington
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Kenneth A. Pass Ph.D.	APHL Liaison
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Marie Mann, M.D.	Department of Health and Human Services Liaison and NNSGRC Project Officer
Susan Panny, M.D.	American College of Medical Genetics Liaison
Jim Eckman, M.D.	Professional Sickle Cell Organizations Liaison
Nathan Bauer, D.V.M.	Parents of Galactosemic Children
Sonya Ross	Sickle cell consumer representative

National Newborn Screening Report - 2000

In order to facilitate and enhance newborn screening data collection efforts, CORN's Newborn Screening Committee assumed responsibility for preparation of the National Newborn Screening Report in October, 1992. A cooperative effort was initiated with APHL's Newborn Screening Committee in order to improve data quality and preclude duplication of effort. This cooperative arrangement has been maintained as the National Newborn Screening Report has continued as a project of the NNSGRC.

Annual National Newborn Screening Reports are designed to respond to the needs of federal, state and local health agencies for pertinent data. It is anticipated that this report will provide data to aid in evaluating the scope and effectiveness of existing state screening programs as well as to assist in identifying unmet needs.

The NNSGRC has not attempted to analyze the data, but has instead concentrated on compilation and presentation of the most current and comprehensive information available. Interpretive comments are included to explain limitations of the compiled information. Interested readers are referred to the document *Newborn Screening - An overview of newborn screening programs in the United States, Canada, Puerto Rico and the Virgin Islands 2000* available from the NNSGRC office. This document contains detailed information about the structure and functions of individual screening programs. It is a valuable source of complementary information.

Data Collection Method

The *National Newborn Screening Report - 2000* is a compilation of 2000 information obtained from state and territorial health departments in response to the NNSGRC's Newborn Screening (NBS) Questionnaire. Surveys were completed by state or territorial personnel identified as the most knowledgeable about their particular newborn screening system.

Data were compiled from the returned questionnaires and a draft report prepared.

Although attempts have been made to make this report as reliable as possible, its accuracy is dependent upon data provided by the individual programs. A listing of responders and reviewers for each screening program is provided in Appendix A of this report.

Participation

Information was solicited from all fifty states, the District of Columbia, Puerto Rico and the Virgin Islands.

Data Collection Period

Data were requested for the period January 1, 2000 through December 31, 2000

Abbreviations

The following is a list of common abbreviations used throughout this report. Explanatory footnotes are provided for individual data tables as needed.

NR	=	No response
N/C	=	Data requested were not collected by the program.
N/A	=	Data were not available in format requested.
N/D	=	No data to report.
DA	=	Does not apply.
~	=	Estimates given by the reporting program.
>	=	Greater than
<	=	Less than
≥	=	Greater than or equal to
≤	=	Less than or equal to

1. Live Birth Statistics

1.0 Introduction

In order to evaluate the effectiveness of the newborn screening program in reaching its target population (all newborns), it is necessary to know the number of live births which have occurred within the program's geographic boundaries. In the past, programs responding to the NBS questionnaire were asked to provide the number of Newborns born in each surveyed geographical area. Responses to this request varied leading to the provision of inconsistent, and at times inaccurate, information. In some cases, responders supplied births to residents regardless of place of occurrence and maintained program data accordingly.

Rather than use self-reported data of variable validity, this report contains tabular live birth data obtained from a central database maintained by the National Center for Health Statistics (NCHS) as reported by state and territorial birth registrars. Data for Puerto Rico and the Virgin Islands were not available and were reported by their respective screening programs. These data are considered provisional for an extended period of time before becoming final and provide the most reliable uniform data set for comparative purpose. Occurrence data is used because screening laws pertain to births occurring within the birth jurisdiction. Resident births are not considered appropriate for comparison with the data collected here.

1.1 Live Births by Occurrence Subdivided by Race

Live birth data reported by place of occurrence and subdivided by race are listed in Table 1.01. These data were obtained from NCHS and will eventually be published as final data in **Vital Statistics of the U.S., Vol. 1, Natality 2000**. The

rates listed are those used by the U.S. Census Bureau. Hispanic origin constitutes a separate category and is further subdivided in Table 1.03.

1.2 Asian Live Birth Data

Table 1.02 lists subdivisions of Asian birth data. These data were also obtained from NCHS and are by place of occurrence.

1.3 Hispanic Live Birth Data

Table 1.03 lists Hispanic births subdivided according to NCHS classifications. Several state newborn screening programs have reported Hispanic birth information as a separate category and are footnoted accordingly.

1.4 Comments

It is important to make the distinction between total births occurring within a geographic location and those occurring only to residents. Because hospitals and medical practitioners within a state or territory are bound by the birth laws of that state or territory, all newborns are screened accordingly without consideration of residence.

In some instances, data are maintained on residents to meet specific legal or other programmatic requirements. Explanations of data maintained in this manner accompany appropriate tables. There is confusion about births within U.S. military or other federal facilities with respect to whether or not they may be governed by local program rules and thus, application of occurrence data as epidemiologic denominators, when assessing incidence, provision of service, etc., may be affected by large federal birthing facilities in the state or territory. Specific information

concerning an individual programs geographic boundaries and the population it serves can be obtained by contacting the individual program in question.

It is also important to note that the vast majority of screening programs do not have a good way of cross-checking specimens received with birth records. Also, there are some federal facilities that may send screening specimens to another state as a result of pricing or other concerns all this data may be included as a part of another state's reporting mechanism.

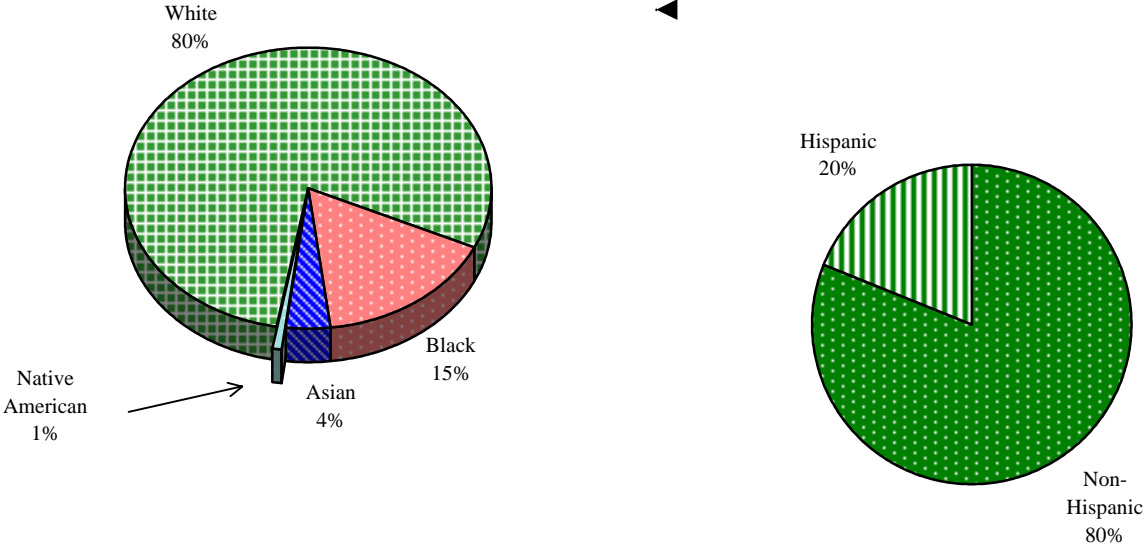
Table 1.01: U.S. Live Births (Occurrence) Classified by Race (2000)*

State/Territory	White	Black	American Indians	Asian or Pacific Is. <i>(See Table 1.02 for Asian Breakdown)</i>	Total	Hispanic <i>(See Table 1.03 for Hispanic Breakdown)</i>	Non-Hispanic
1 Alabama	41,606	20,246	173	537	62,562	1,934	60,628
2 Alaska	6,265	461	2,507	633	9,866	948	8,918
3 Arizona	75,061	2,803	5,539	2,067	85,470	36,415	49,055
4 Arkansas	28,664	7,405	271	500	36,840	2,438	34,402
5 California	430,283	35,037	3,023	64,267	532,610	261,953	270,657
6 Colorado	59,927	3,034	642	2,076	65,679	18,305	47,374
7 Connecticut	36,123	5,277	127	1,843	43,370	7,510	35,860
8 Delaware	8,634	2,596	39	370	11,639	1,049	10,590
9 District of Columbia	5,783	8,840	14	522	15,159	1,359	13,800
10 Florida	150,692	47,457	1,134	5,022	204,305	46,189	158,116
11 Georgia	85,138	44,535	302	3,549	133,524	15,211	118,313
12 Hawaii	4,043	475	189	12,931	17,638	2,393	15,245
13 Idaho	19,218	79	272	294	19,863	2,631	17,232
14 Illinois	139,672	34,006	270	8,036	181,984	39,304	142,680
15 Indiana	77,022	9,527	142	1,200	87,891	5,931	81,960
16 Iowa	35,886	1,243	364	925	38,418	2,519	35,899
17 Kansas	34,810	2,969	411	1,042	39,232	5,196	34,036
18 Kentucky	48,851	4,934	67	571	54,423	1,046	53,377
19 Louisiana	38,607	28,242	390	1,036	68,275	1,697	66,578
20 Maine	13,042	115	119	186	13,462	157	13,305
21 Maryland	44,609	21,240	237	3,488	69,574	4,732	64,842
22 Massachusetts	69,559	8,119	164	4,831	82,673	9,801	72,872
23 Michigan	106,127	24,277	672	3,813	134,889	15,642	119,247
24 Minnesota	58,392	4,447	1,217	3,490	67,546	6,378	61,168
25 Mississippi	22,322	20,028	246	384	42,980	658	42,322
26 Missouri	64,790	11,625	363	1,524	78,302	2,697	75,605
27 Montana	9,448	45	1,321	113	10,927	700	10,227
28 Nebraska	22,623	1,374	393	571	24,961	2,972	21,989
29 Nevada	25,600	2,372	428	1,987	30,387	10,362	20,025
30 New Hampshire	13,492	161	31	303	13,987	1,070	12,917
31 New Jersey	82,170	20,880	178	9,083	112,311	22,653	89,658
32 New Mexico	22,271	479	3,658	401	26,809	13,520	13,289
33 New York	184,362	55,038	714	19,881	259,995	65,963	194,032
34 North Carolina	87,263	29,568	1,731	2,785	121,347	12,658	108,689
35 North Dakota	7,743	87	913	104	8,847	517	8,330
36 Ohio	128,934	23,756	330	2,923	155,943	4,505	151,438
37 Oklahoma	37,713	4,766	5,189	982	48,650	4,871	43,779
38 Oregon	42,600	1,053	737	2,400	46,790	7,659	39,131
39 Pennsylvania	121,663	20,801	391	4,002	146,857	8,278	138,579
40 Rhode Island	11,451	1,115	159	455	13,180	3,194	9,986
41 South Carolina	33,319	19,211	197	835	53,562	2,274	51,288
42 South Dakota	8,861	106	1,495	127	10,589	247	10,342
43 Tennessee	65,637	17,662	172	1,361	84,832	3,401	81,431
44 Texas	313,908	41,543	825	11,743	368,019	171,760	196,259
45 Utah	46,017	371	619	1,447	48,454	6,308	42,146
46 Vermont	6,159	28	13	77	6,277	107	6,170
47 Virginia	69,249	22,492	105	4,909	96,755	7,810	88,945
48 Washington	68,156	3,462	1,990	6,845	80,453	13,726	66,727
49 West Virginia	20,709	796	10	105	21,620	124	21,496
50 Wisconsin	58,726	6,505	930	2,089	68,250	4,504	63,746
51 Wyoming	5,473	55	262	57	5,847	546	5,301
52 Puerto Rico a					0	59,461	
53 Virgin Islands b	74	1,406		37	1,851	334	1,517
TOTAL	3,198,747	624,149	41,685	200,759	4,065,674	923,617	3,142,057

* = Unpublished data supplied by NCHS excluding Puerto Rico and the Virgin Islands.

a = data reported by Puerto Rico; b = data reported by Virgin Islands.

Figure 1.01: U.S. Live Births (Occurrence) Classified by Race for 2000



See Non-Hispanic
vs. Hispanic
Table 1.01

Table 1.02: Asian Live Births by Occurrence in the U.S. (2000)*

State/Territory	Chinese	Japanese	Hawaiian	Filipino	Other Asian P. Islander	Total Asian
1 Alabama	91	24	4	48	370	537
2 Alaska	15	19	21	283	295	633
3 Arizona	173	57	44	263	1,530	2,067
4 Arkansas	52	15	5	59	369	500
5 California	13,615	3,087	793	15,315	31,457	64,267
6 Colorado	194	75	36	154	1,617	2,076
7 Connecticut	311	87	5	167	1,273	1,843
8 Delaware	40	1	3	29	297	370
9 District of Columbia	96	29	2	113	282	522
10 Florida	485	161	34	975	3,367	5,022
11 Georgia	160	60	35	168	3,126	3,549
12 Hawaii	674	2,151	4,719	3,512	1,875	12,931
13 Idaho	33	43	13	42	163	294
14 Illinois	1,056	251	15	1,518	5,196	8,036
15 Indiana	124	50	16	94	916	1,200
16 Iowa	128	17	16	67	697	925
17 Kansas	62	8	7	30	935	1,042
18 Kentucky	50	38	5	36	442	571
19 Louisiana	74	18	9	71	864	1,036
20 Maine	24	6	3	33	120	186
21 Maryland	454	85	10	304	2,635	3,488
22 Massachusetts	1,293	184	7	194	3,153	4,831
23 Michigan	499	146	22	321	2,825	3,813
24 Minnesota	206	51	14	122	3,097	3,490
25 Mississippi	24	3	2	30	325	384
26 Missouri	229	51	22	200	1,022	1,524
27 Montana	13	17	8	28	47	113
28 Nebraska	48	19	9	59	436	571
29 Nevada	213	115	119	782	758	1,987
30 New Hampshire	60	15	6	31	191	303
31 New Jersey	1,633	246	16	1,412	5,776	9,083
32 New Mexico	60	25	7	68	241	401
33 New York	7,613	593	25	1,373	10,277	19,881
34 North Carolina	183	69	19	172	2,342	2,785
35 North Dakota	10	5	0	13	76	104
36 Ohio	293	96	12	158	2,364	2,923
37 Oklahoma	83	20	18	45	816	982
38 Oregon	286	147	48	240	1,679	2,400
39 Pennsylvania	650	95	12	160	3,085	4,002
40 Rhode Island	2	0	0	9	444	455
41 South Carolina	87	35	8	90	615	835
42 South Dakota	12	2	0	24	89	127
43 Tennessee	86	30	9	83	1,153	1,361
44 Texas	1,464	254	97	1,204	8,724	11,743
45 Utah	95	64	47	58	1,183	1,447
46 Vermont	15	4	1	4	53	77
47 Virginia	480	101	18	714	3,596	4,909
48 Washington	542	284	258	1,096	4,665	6,845
49 West Virginia	12	5	2	11	75	105
50 Wisconsin	192	46	5	129	1,717	2,089
51 Wyoming	4	0	4	15	34	57
52 Puerto Rico						
53 Virgin Islands a					37	37
TOTAL	34,298	9,004	6,610	32,126	118,721	200,759

* = Unpublished data supplied by NCHS excluding Puerto Rico and the Virgin Islands.

a = data reported by Virgin Islands.

Figure 1.02: Asian Live Births by Occurrence in the U.S. for 2000

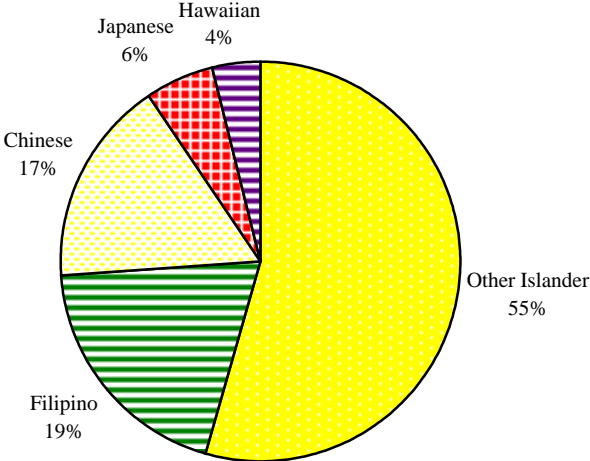


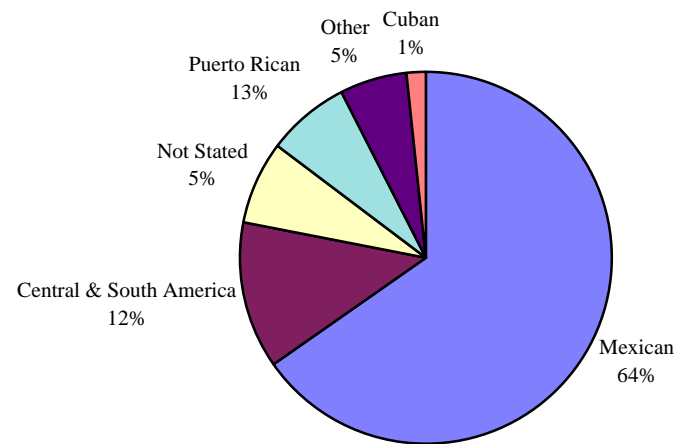
Table 1.03: Hispanic Live Births by Occurrence in the U.S. (2000)*

State/Territory	Mexican	Puerto Rican	Cuban	Central and South America	Other	Not Stated	Total Hispanic
1 Alabama	1,406	81	22	157	225	43	1,934
2 Alaska	266	48	9	64	203	358	948
3 Arizona	33,175	241	56	641	601	1,701	36,415
4 Arkansas	1,949	37	8	285	58	101	2,438
5 California	225,571	2,073	733	25,134	4,528	3,914	261,953
6 Colorado	13,641	210	35	621	3,752	46	18,305
7 Connecticut	591	4,122	78	1,536	209	974	7,510
8 Delaware	498	327	9	186	12	17	1,049
9 District of Columbia	140	14	9	1,102	44	50	1,359
10 Florida	10,814	8,459	9,316	15,964	1,253	383	46,189
11 Georgia	10,491	512	157	2,107	121	1,823	15,211
12 Hawaii	451	750	9	100	991	92	2,393
13 Idaho	2,162	15	5	62	273	114	2,631
14 Illinois	33,121	2,884	181	1,616	1,408	94	39,304
15 Indiana	4,706	323	32	382	100	388	5,931
16 Iowa	1,808	43	10	299	124	235	2,519
17 Kansas	3,949	94	16	238	432	467	5,196
18 Kentucky	728	70	41	146	11	50	1,046
19 Louisiana	695	101	62	141	559	139	1,697
20 Maine	29	24	1	31	50	22	157
21 Maryland	909	344	51	2,387	794	247	4,732
22 Massachusetts	391	4,545	79	3,957	328	501	9,801
23 Michigan	5,520	437	79	395	460	8,751	15,642
24 Minnesota	3,109	111	27	437	199	2,495	6,378
25 Mississippi	340	23	5	44	203	43	658
26 Missouri	1,911	119	47	409	176	35	2,697
27 Montana	154	9	4	16	146	371	700
28 Nebraska	1,954	24	8	320	151	515	2,972
29 Nevada	8,245	161	176	926	533	321	10,362
30 New Hampshire	99	78	4	122	55	712	1,070
31 New Jersey	3,436	6,984	858	10,588	312	475	22,653
32 New Mexico	4,701	51	33	129	8,570	36	13,520
33 New York	8,045	14,086	476	22,442	8,995	11,919	65,963
34 North Carolina	9,524	621	105	2,209	109	90	12,658
35 North Dakota	166	7	0	11	32	301	517
36 Ohio	2,197	1,331	45	440	188	304	4,505
37 Oklahoma	3,336	104	18	217	651	545	4,871
38 Oregon	6,937	91	42	315	147	127	7,659
39 Pennsylvania	1,294	4,931	91	521	727	714	8,278
40 Rhode Island	130	648	14	1,233	86	1,083	3,194
41 South Carolina	1,565	131	18	332	155	73	2,274
42 South Dakota	149	14	0	57	23	4	247
43 Tennessee	2,483	214	42	497	123	42	3,401
44 Texas	149,126	1,071	270	8,367	8,598	4,328	171,760
45 Utah	4,841	100	14	560	593	200	6,308
46 Vermont	15	13	3	3	4	69	107
47 Virginia	1,769	558	57	4,750	493	183	7,810
48 Washington	9,431	268	40	596	1,036	2,355	13,726
49 West Virginia	36	1	2	3	8	74	124
50 Wisconsin	3,425	624	32	248	162	13	4,504
51 Wyoming	486	6	0	1	45	8	546
52 Puerto Rico a		59,000	100	61	300		59,461
53 Virgin Islands b					334		334
TOTAL	581,915	117,133	13,529	113,405	49,690	47,945	923,617

* = Unpublished data supplied by NCHS excluding Puerto Rico and the Virgin Islands.

a = data reported by Puerto Rico; **b** = data reported by Virgin Islands.

Figure 1.03: Hispanic Live Births by Occurrence in the U.S. for 2000



2. Newborn Screening Overview

2.0 Introduction

It is extremely difficult to compare data from various screening programs without the use of standard definitions. As screening has progressed, so has our understanding of the disorders involved. In most instances, screening has led to the discovery of a multitude of manifestations of what previously were considered to be simple disorders. Correspondingly, the definitions of degrees of severity of a disorder, and their relationship to laboratory screening results, determine the actions of both laboratory and follow-up personnel. Consensus definitions have not yet been reached among the various screening programs. In order to try to arrive at such definitions, programs were once again asked to share their definitions of various disorders.

2.1 Disorders Included in Newborn Screening Programs - 2000

Table 2.01 lists each newborn screening program and the disorders included for testing in 2000. Most programs were guided by statutes or rules requiring that certain screening tests be performed. When testing was required, the letter "R" is used in the table. In non-required (or voluntary) screening situations, the letter "V" is used.

2.2 Laboratories Providing Newborn Screening Tests

Table 2.02 provides a summation of the type of laboratory service(s) available. Programs were asked to respond with information denoting the type of laboratory service(s) available. While

most programs used a single laboratory, others permitted testing at one or more laboratories within their state or territory. Some programs contract for service from a laboratory outside of their geographic boundaries.

2.3 Components Included in Newborn Screening Follow-Up

Follow-up is a complex issue and is usually approached on several levels (e.g., telephone contact, certified mail, personal visits, etc.) depending on the size of the program, its resources and the anticipated outcome to the patient based on the screening laboratory's results. In order to provide a comparison of follow-up systems, programs were asked to indicate the type(s) of follow-up utilized for pursuing abnormal screening results. This information is displayed in Table 2.03. While all programs reported notifying submitters of abnormal laboratory test results, several did not indicate a formal program of confirmation and treatment, particularly in states with multiple or regional laboratories. Long term follow-up of diagnosed cases was less uniform and some programs indicated a lack of significant follow-up beyond the initial diagnostic and treatment phase.

2.4 Age at Time of Newborn Screening

The use of biochemical markers for detecting genetic and metabolic disorders is dependent on their quantitative levels at the time of screening.

Some disorders are more difficult to detect using samples collected close to the time of birth. Since collection of the initial screening sample is recommended before hospital discharge, it is important to monitor trends in specimen collection, given the national trend towards early hospital discharge for maternity patients.

Table 2.04 lists information from each program relative to the newborn's age at time of sample collection. Not all programs obtain specific time of sample collection and thus estimates were reported in some instances. *These data refer only to initial samples and do not contain information relative to requested or required follow-up after initial testing.*

2.5 Summation of Fees Charged for Newborn Screening

Over the years it has become necessary for programs to consider the initiation of fees as a means of recovering expenses. In some cases these fees cover only the laboratory testing expenses, while in other cases, there are significant other expenses included in the fee calculations. There are also significant differences between programs as to how the fee is charged and how the monies are made available within the program. Table 2.05 lists the answers to the basic questions of the amount and coverage of the fee. If this information is considered useful and other information should be requested for tabulation, please make this suggestion to the editors.

2.6 Criteria for Secondary Screening Tests

Most U.S. newborn screening programs include certain components of follow-up that result in a

second filter paper specimen being tested. Additionally, some programs require second testing on all newborns at some period after the baby is discharged from the hospital. Some programs also require an automatic second screen if the first is taken at a time defined as 'too early' by the program. In order to better define the circumstances that result in submission of a second screening sample, programs were asked to submit the information tabulated in Table 2.06.

2.7 Laboratory Specimen Information

Programs were asked to submit certain summary information about their screening programs, sample quality, educational programs, and sample storage policies. These data were accumulated for comparison of program efforts at improving sample quality and to ascertain policy developments relative to sample storage and use. Additionally, information about evaluation of sample quality, computer approaches to quality improvement, and educational emphasis were requested. These data are summarized in Table 2.07.

2.8 Comments

In many cases, definitions of a disorder were essentially the same throughout all programs. In others this was not the case. It is a national goal to arrive at standardized definitions, action levels, and actions for the various screening disorders. More specific definitions are necessary in order to make the data submitted more meaningful. Only programs screening for the disorder listed are included in the tables that follow.

Table 2.01: Disorders for Which Newborns Were Screened in the U.S. (2000)

State/Territory	Hyperphenylalaninemia	Hypothyroidism	Classical Galactosemia	Maple Syrup Urine Disease	Homocystinuria	Biotinidase	Congenital Adrenal Hyperplasia	Cystic Fibrosis	Tyrosinemia	Toxoplasmosis	Hemoglobinopathy	MCAD
1 Alabama	R	R	R				R				R	
2 Alaska	R	R	R	R		R	R					
3 Arizona	R	R	R	R	R	R					R	
4 Arkansas	R	R	R								R	
5 California	R	R	R								R	
6 Colorado	R	R	R			R	R b	R			R	
7 Connecticut	R	R	R	R	R	R	R	V			R	
8 Delaware	R	R	R								R	
9 District of Columbia	R	R	R	R	R						R	
10 Florida	R	R	R				R				R	
11 Georgia	R	R	R	R	R		R		R		R	
12 Hawaii	R	R	R	R		R	R				R	
13 Idaho	R	R	R	R		R						
14 Illinois	R	R	R a			R	R				R	
15 Indiana	R	R	R	R	R	R	R				R	
16 Iowa	R	R	R				R				R	P
17 Kansas	R	R	R								R	
18 Kentucky	R	R	R								V	
19 Louisiana	R	R				R					R	
20 Maine	R	R	R	R	R	R	R				V	R
21 Maryland d	V	V	V	V	V	V			V		V	
22 Massachusetts	R	R	R	R	R	R	R	P	P	R	R	R
23 Michigan	R	R	R	R		R	R				R	
24 Minnesota	R	R	R				R				R	
25 Mississippi	R	R	R								R	
26 Missouri	R	R	R								R	
27 Montana	R	R	R					V			P	
28 Nebraska	R	R	R			R					R	
29 Nevada	R	R	R	R		R					R	
30 New Hampshire	R	R	R	R	R					R	V	
31 New Jersey	R	R	R								R	
32 New Mexico	R	R	R			R	R				R	
33 New York e	R	R	R	R	R	R					R	
34 North Carolina	R	R	R	R	R		R		R		R	R
35 North Dakota	R	R	R				R				V	
36 Ohio	R	R	R		R						R	
37 Oklahoma	R	R	R								R	
38 Oregon	R	R	R	R		R					R	
39 Pennsylvania	R	R	V & R f	R	V a	V a	V & R f	V a			R	
40 Rhode Island	R	R	R	R	R	R	R				R	
41 South Carolina	R	R	R				R				R	R
42 South Dakota	R	R	R									
43 Tennessee	R	R	R				R c				R	
44 Texas	R	R	R				R				R	
45 Utah	R	R	R									
46 Vermont	R	R	R	R	R	R					R	
47 Virginia	R	R	R	R	R	R					R	
48 Washington	R	R					R				R	
49 West Virginia	R	R	R								V	
50 Wisconsin	R	R	R			R	R	R			R	
51 Wyoming	R	R	R			R		R			R	
52 Puerto Rico	R	R	P								R	
53 Virgin Islands	R	R	R	R	R						R	

R = Required; V = Voluntary; P = Pilot

a = supplemental newborn screening program through Neo Gen. Screening, Inc.; b = CAH testing since 8/00; c = started screening 10/00; d = Maryland law requires that hospitals or birthing centers must offer testing, which can be refused without reason; therefore, for purposes of this report, Maryland is listed as voluntary; e = other disorders screened HIV-1 antibodies;

f = supplemental newborn screening program through Neo Gen. Screening, Inc. (Voluntary 1/1/00 to 9/31/00 and part of PA state screening program 10/1/00 to 12/31/00.

Figure 2.01: Disorders Screened in 2000

All states screen for Hyperphenylalaninemia and Hypothyroidism

Totals for other disorders may be inaccurate due to some states not reporting

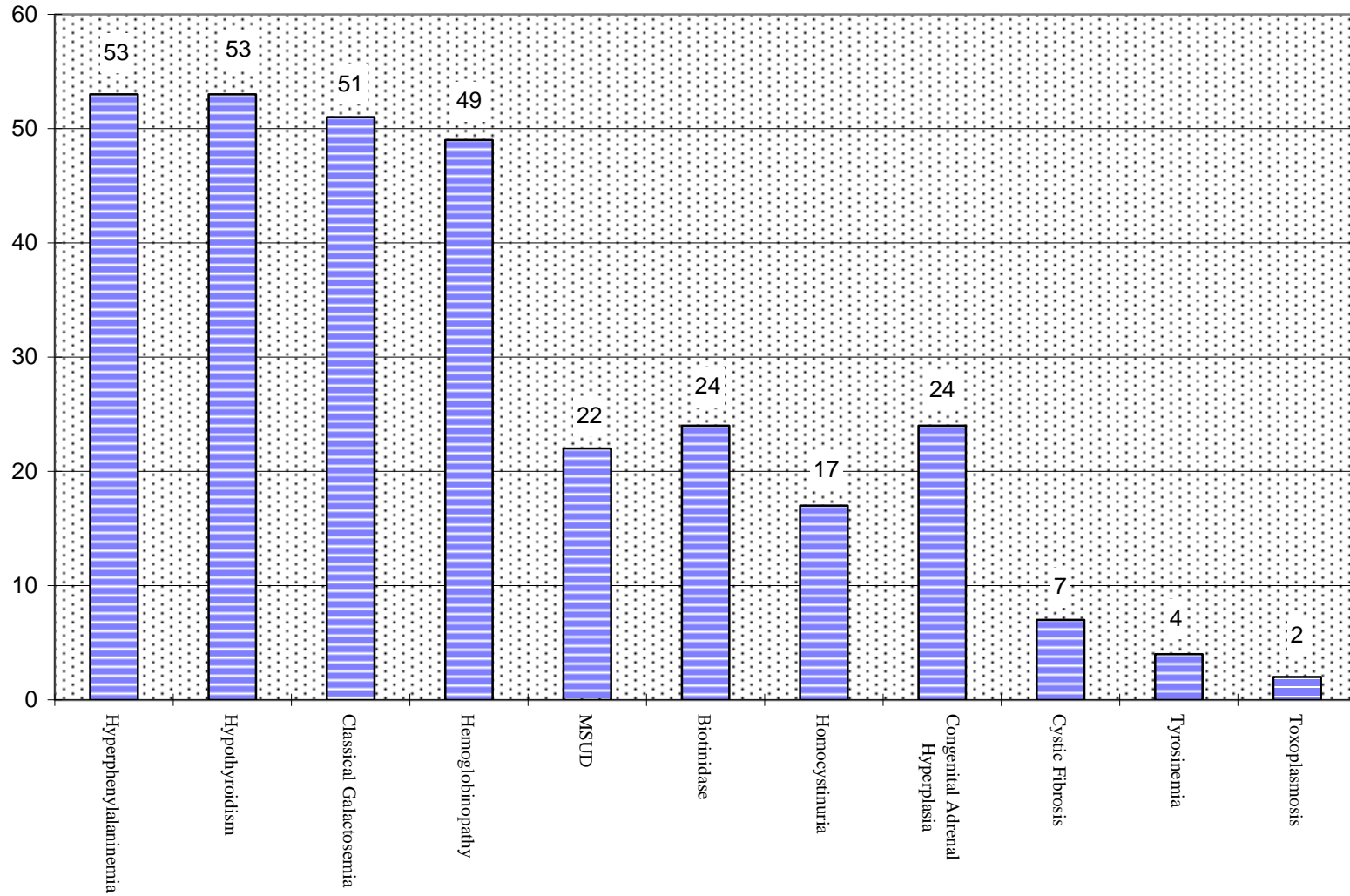


Table 2.02: Summation of Laboratories Providing Newborns Screening Services in the U.S.

State/Territory	Laboratories Operated by State	Using Regional Lab	Private Labs Under State Regulations	Private Labs Without State Regulation	Total Number of Labs Testing
1 Alabama	1				1
2 Alaska		Contracts w/Oregon			1
3 Arizona	1				1
4 Arkansas	1				1
5 California	1		8		9
6 Colorado	1				1
7 Connecticut	1				1
8 Delaware	1				1
9 District of Columbia			1		1
10 Florida	1				1
11 Georgia	1				1
12 Hawaii		Contracts w/Oregon			1
13 Idaho		Contracts w/Oregon			1
14 Illinois	1				1
15 Indiana			1		1
16 Iowa	1				1
17 Kansas	1				1
18 Kentucky	1	DA	2		3
19 Louisiana	1			2	3
20 Maine		1			1
21 Maryland	1				1
22 Massachusetts	1				1
23 Michigan	1				1
24 Minnesota	1				1
25 Mississippi		1			1
26 Missouri	1				1
27 Montana	1				1
28 Nebraska a			3		3
29 Nevada		Contracts w/Oregon			1
30 New Hampshire		Contracts w/Mass.			1
31 New Jersey	1				1
32 New Mexico	1				1
33 New York	1				1
34 North Carolina	1				1
35 North Dakota		Contracts w/Iowa			1
36 Ohio	1				1
37 Oklahoma	1				1
38 Oregon	1				1
39 Pennsylvania			1	1 b	2
40 Rhode Island		Contracts w/Mass.			1
41 South Carolina	1				1
42 South Dakota				1	1
43 Tennessee	1				1
44 Texas	1				1
45 Utah	1				1
46 Vermont		Contracts w/Mass.			1
47 Virginia	1				1
48 Washington	1				1
49 West Virginia	1				1
50 Wisconsin	1				1
51 Wyoming		Contracts w/Colorado			1
52 Puerto Rico		1	1		2
53 Virgin Islands			1		1

a = 3 NBS labs in NE through 2-29-00, after only two, Colorado lab conducts NBS tests for births at Federal Military Hosp, 509 screened at Colorado lab; **b** = NeoGen Screening, Inc.

Table 2.03: Components Included in Newborn Screening Follow-up in the U.S.

State/Territory	Notification by Lab Report, Letter and Phone Call	Confirmation of Additional Lab Results	Confirmation of Treatment	Annual Follow-up of Diagnosed Cases	Number of Years Followed
1 Alabama	X	X			
2 Alaska	X	X	X		
3 Arizona	X	X	X		AZ follows baby until final dx & treatment, then followed by other programs.
4 Arkansas	X	X	X	X	Up to 5 years of age.
5 California	X	X	X		Maternal PKU females ≥13 years.
6 Colorado	X	X	X		
7 Connecticut	X	X	X		Maintain permanent files on all confirmed disease cases.
8 Delaware	X	X	X	X	Annual follow-up some cases especially PKU. Years followed variable.
9 District of Columbia	X	X			The lab notifies the submitter & DC MCH who is responsible for follow-up.
10 Florida	X	X	X		
11 Georgia	X	X	X	X	Indefinite
12 Hawaii	X	X	X	X	Diagnosed cases followed annually up to 21 yrs. of age, if admitted to Children w/Special Health Needs Branch.
13 Idaho	X	X	X	X	18 + years
14 Illinois	X	X	X	X	15 years
15 Indiana	X a	X a	X b		
16 Iowa	X	X	X	X	Lifetime, some follow-up are followed out of state.
17 Kansas	X	X	X	X	Birth to death if they are a client of Special Health Services.
18 Kentucky	X	X	X	X	Varies
19 Louisiana	X	X	X	X	PKU patients followed for life, congenital hypothyroidism tracked to verification of trmt., SC followed for 5 years.
20 Maine	X	X	X		
21 Maryland	X	X	X	X	Hgb - 3 years; Metabolic - indefinite as long as on diet.
22 Massachusetts	X	X	X	X	Varies by disorder
23 Michigan	X	X	X	X	21 years
24 Minnesota	X	X	X	X	
25 Mississippi	X	X	X	X	21 years
26 Missouri	X				
27 Montana	X	X	X	X	19 years
28 Nebraska	X	X	X	X	Followed at treatment center for Galactosemia, Biotinidase Deficiency and Phenylketonuria (PKU).
29 Nevada	X	X	X	X	21 years, all abnormal referred to Early Intervention Clinics.
30 New Hampshire	X	X	X	X	Varies by disorder, Diagnosed cases up to 21 years.
31 New Jersey	X	X	X	X (SCD only)	1, 2, 5, 10, 15 and 20 years.
32 New Mexico	X	X	X	No	1 year
33 New York	X	X			NY State conducts follow-up activities until a diagnosis is received.
34 North Carolina	X	DA	X	DA	
35 North Dakota	X	X	X	X	21 years, Title V provides formula for PKU & MSUD patients.
36 Ohio	X	X	X		
37 Oklahoma	X	X	X	X	6 years, Sickle cell disease and other Hgbs followed for six years.
38 Oregon	X	X	X		
39 Pennsylvania	X	X	X	X	21 years by the treatment centers, annual follow-up by treatment ctrs.
40 Rhode Island	X	X	X	X	indefinite
41 South Carolina	X	X	X	X	Followed indefinitely for PKU only.
42 South Dakota	X	X	X		
43 Tennessee	X	X	X		
44 Texas	X	X	X	X	≥ 17 years, outreach & case management prog., and comm. resources.
45 Utah	X	X	X		
46 Vermont	X	X	X	X	Varies by condition.
47 Virginia	X	X	X	X	Annual follow-up is handled by the metabolic specialist.
48 Washington	X	X	X	X	Varies by disorder. c
49 West Virginia	X	X	X	X	Varies by disorder.
50 Wisconsin	X	X	X		
51 Wyoming	X	X	X		
52 Puerto Rico	X	X	X	X	18 yrs, few cases referred to local specialists.
53 Virgin Islands	X	X			

a = services provided by contract lab; **b** = services provided by state (MCH); **c** = follow-up of infants not screened & with unsat specimens.

Table 2.04: Infant's Age at Time of Initial Testing in U.S. Newborn Screening Programs

State/Territory	0-12	13-24	Day 1 (Total of 1st two columns)	Day 2 25-48	Day 3 49-72	Day 4 73-96	Day 5 97-120	Day 6 121-144	Day 7 145-168	Over 7 Days 169-999	Hour Unknown	Calculated TOTAL	
1 Alabama											62,173	62,173	
2 Alaska	832	1,698	2,530	3,982	1,495	537	154	254	100	277	492	9,821	
3 Arizona	1,068	12,174	13,242	44,817	14,544	4,285	1,062	541	20	1,259	2,642 g	81,956	
4 Arkansas	814	1,111	1,925	29,001	3,087	830	391	366	268	695	139	36,446	
5 California	14,123	160,329	174,452	263,044	47,934	15,365	8,375	5,663	2,303	4,618	5,543	527,297	
6 Colorado											63,219	63,219	
7 Connecticut											43,723	43,723	
8 Delaware	88	639	727	7,495	2,131	409	889			63	0	11,714	
9 District of Columbia											15,125	15,125	
10 Florida											204,030	204,030	
11 Georgia											189,498	189,498	
12 Hawaii	49	1,767	1,816	12,953	2,193	433	36	31	26	112	12	17,612	
13 Idaho	1,089	4,294	5,383	8,326	3,213	1,528	354	189	102	613	546	20,254	
14 Illinois d	209	720	929	6,999	1,279	3,382					175,575	188,164	
15 Indiana	2,164	4,926	7,090	21,469	51,561	3,440	1,506	885	580	1,071	37	87,639	
16 Iowa	87	302	389	31,404	4,864	793	232	63	36	229	131	38,141	
17 Kansas	759	864	1,623	28,017	6,567	1,039	350	740	312	382	1	39,031	
18 Kentucky											54,515		
19 Louisiana											67,843	67,843	
20 Maine	N/A	N/A	2,767	<----- 10,394 ----->			<--- 101 --->		<--- 79 ----->			13,341	
21 Maryland			19,489	38,474	5,045	1,613	708	609	561	2,667	3,224	72,390	
22 Massachusetts			1,285	48,681	22,539	4,724	739	133	71	525 f	4,006	82,703	
23 Michigan	1,588	31,142	32,730	85,131	5,493	1,055	320	159	109	878	8,147	134,022	
24 Minnesota	209	3,026	3,235	50,999	9,953	1,208	436	175	74	868	1,454	68,402	
25 Mississippi											44,075	44,075	
26 Missouri	573	2,251	2,824	50,027	15,110	4,070	1,231	1,017	1,017	2,055	1,289	78,640	
27 Montana			805								10,020	10,825	
28 Nebraska											24,863	24,863	
29 Nevada	3,323	11,075	14,398	10,798	2,749	807	101	83	69	600	1,054	30,659	
30 New Hampshire			100	9,769	3,055	461	83	71	59	91 a	4,010	13,879	
31 New Jersey	723	644	1,367	91,305	17,174	1,113	308	115	78	467	314	112,241	
32 New Mexico			24,795	61	12	4		5	2	16	970	25,865	
33 New York	N/A	N/A	1,437	42,221	163,981	32,961	10,384	3,767	967	2,343	388	258,449	
34 North Carolina	781	15,134	15,915	89,460	10,939	1,550	533	367	219	1,028	739	120,750	
35 North Dakota	72	78	150	6,745	1,427	278	77	23	16	44	46	8,806	
36 Ohio											160,566	160,566	
37 Oklahoma			N/C								52,760	52,760	
38 Oregon	2,740	8,699	11,439	23,138	6,620	1,762	165	112	76	423	3,144	46,879	
39 Pennsylvania e											148,597	148,597	
40 Rhode Island			5,163	<----- 7,824 ----->			<--- 68 --->		<--- 98 ----->			13,150	
41 South Carolina		749	<----->					52,475		<----->		396	52,176
42 South Dakota											10,760	10,760	
43 Tennessee											79,539	79,539	
44 Texas											355,100	355,100	
45 Utah											47,423	47,423	
46 Vermont	25	83	108	4,854	568	174	201	63	16	50	6	6,040	
47 Virginia			14,577	68,958	9,051	1,507	799	588	698	2,941	291	99,410	
48 Washington	9,379	37,075	46,454	19,239	5,939	1,690	873	260	81	791	1,559	76,886 b	
49 West Virginia	46	3,691	3,737	11,523	3,419	727	413	283	543	339	337	21,321	
50 Wisconsin	392	1,594	1,986	49,769	11,584	2,015	519	531	384	636	173	66,614	
51 Wyoming											5,480	5,480	
52 Puerto Rico		30	30 c	57,075						50		57,155	
53 Virgin Islands			370	1,481								1,851	

a = includes out of state adoptions; b = excludes 2,928 military births & 3,207 exclusions, 55 others screened by OR, 144 neonatal deaths & 80 refusals; c = estimated number; d = data for abnormal results only; e = supplemental newborn screening program through NeoGen screening has data in different format, approximately 5.5% of specimens received were collected at < 24 hours of age; f = includes out of state adoptions; g = of DOB, time of birth, date of collection or time of collection were blank counted as "Exact Time Unknown".

Table 2.05: Summation of Fees Charged in 2000 for Newborn Screening

State/Territory	Amount of Fee	Program Components covered by fee
1 Alabama	\$24.00	Laboratory
2 Alaska	\$24.00	Laboratory, Program Administration/Follow-up
3 Arizona	\$20 / \$15	Laboratory, Program Administration/Follow-up, Treatment, specialist consultation, nurses
4 Arkansas	\$14.83	Laboratory
5 California	\$42.00	Laboratory, Program Administration/Follow-up
6 Colorado	\$33.50	Laboratory, Program Administration/Follow-up, Treatment and Genetic Counseling
7 Connecticut a	\$18.00	Laboratory
8 Delaware	\$40.69	Laboratory, Program Administration/Follow-up, Medical Consultant.
9 District of Columbia	No charge	
10 Florida d	\$20.00	Laboratory, Program Administration/Follow-up
11 Georgia	No Charge	
12 Hawaii	\$27.00	Laboratory, Program Administration/Follow-up, Treatment, fed ex, education, consultants/genetics
13 Idaho	No Charge	
14 Illinois	\$32.00	Laboratory, Program Administration/Follow-up, Treatment
15 Indiana	\$28.50	Laboratory, Program Administration/Follow-up and Treatment
16 Iowa	\$33.00	Laboratory, Program Administration/Follow-up
17 Kansas	No Charge	
18 Kentucky	\$14.50	Laboratory
19 Louisiana	\$18.00	Laboratory, Program Administration/Follow-up and Treatment, Surveillance, Education.
20 Maine	\$26.75	Laboratory, Program administration/Follow-up and education.
21 Maryland	\$15.75	Laboratory (reagents only).
22 Massachusetts	\$49.55	Laboratory, Program Administration/Follow-up, in home trait counseling
23 Michigan	\$39.00	Laboratory, Program Administration/Follow-up and some treatment.
24 Minnesota	\$21.00	Laboratory, Program Administration/Follow-up
25 Mississippi	\$35.00	Laboratory, Program Administration/Follow-up and Treatment
26 Missouri	\$13.00	Laboratory
27 Montana	\$36.92 e	Laboratory
28 Nebraska	\$53.00-\$54.60	Laboratory, Treatment
29 Nevada	\$30.00	Laboratory, Program administration/Follow-up
30 New Hampshire	\$18.00	Laboratory
31 New Jersey	\$34.00	Laboratory, Program Administration/Follow-up and Treatment
32 New Mexico	\$20.00	Laboratory, Program Administration/Follow-up and Treatment, Education & Genetic Serv
33 New York	No Charge	
34 North Carolina	No Charge	
35 North Dakota	\$17.00 c	Laboratory
36 Ohio	\$27.00	Laboratory, Program administration/Follow-up, Treatment
37 Oklahoma	\$10.50	Laboratory
38 Oregon	\$32.00	Laboratory, Program Administration/Follow-up, Treatment
39 Pennsylvania		
40 Rhode Island	\$59.00	Laboratory, Program Administration/Follow-up, Specialty formulas.
41 South Carolina	\$21.00	Laboratory and Treatment
42 South Dakota	No Charge	
43 Tennessee	\$17.50 b	Laboratory
44 Texas	\$13.75	Laboratory
45 Utah	\$27.00	Laboratory, Program Administration/Follow-up
46 Vermont	\$27.00	Laboratory, Program Administration/Follow-up
47 Virginia	\$16.00	Laboratory, Program Administration/Follow-up and Treatment, metabolic formula.
48 Washington	\$39.25	Laboratory, Program Administration/follow-up, Treatment, Program evaluation and educ.
49 West Virginia	\$20.46	Laboratory
50 Wisconsin	\$55.50	Laboratory, Program Administration/Follow-up and Treatment.
51 Wyoming	No charge	
52 Puerto Rico	\$18.00	Laboratory, Program Administration/Follow-up, Treatment.
53 Virgin Islands	No charge	
54 NeoGen Screening	19.75	Laboratory, Program Administration/Follow-up

a = Hosp. bill each infant tested, pass charge to insurance co. as part of mty fee; fees cover testing through state lab; **b** = increased to \$17.50 10/00;

c = 1/2000 to 6/2000 fee \$16.00 - 7/2000 to 12/2000 fee \$17.00; **d** = charge based on the number of live births occurring during the previous calendar year;

e = Jan-June, 2000 fee \$35.50, July-Dec, 2000 fee \$36.92.

Table 2.06: Criteria for Second Screening Tests in U.S. Newborn Screening Programs

State/Territory	Required Second Screening on all	Only If Tested Prior to 24 Hours	Only If Tested Prior to 36 Hours	Only If Tested Prior to 48 Hours	Definitions of Selected Groups Receiving 2nd Screening Tests
1 Alabama		X			Second testing recommended on all infants 2-6 weeks of age.
2 Alaska				X	
3 Arizona		X			Recommended for all babies
4 Arkansas		X			
5 California					N/A
6 Colorado	X				
7 Connecticut					Between 7-14 days; all 2nd specimens tested for PKU.
8 Delaware	X				
9 District of Columbia		X			
10 Florida				X	
11 Georgia				X	Low birth weight
12 Hawaii		X			
13 Idaho				X	
14 Illinois		X			
15 Indiana				X	
16 Iowa		X			
17 Kansas		X			
18 Kentucky				X	
19 Louisiana				X	
20 Maine		X			
21 Maryland	X	X			Maryland is a voluntary newborn screening program and all testing is recommended.
22 Massachusetts		X			Retest VLBW (<1500g) & NICU infants-retest at 2 wks of age.
23 Michigan		X			Recommended if specimen sample > 24 hours of age and < 36 hours.
24 Minnesota		X			Required if specimen collected < 24 hours.
25 Mississippi		X			
26 Missouri		X			
27 Montana		X			
28 Nebraska		X			
29 Nevada	X				
30 New Hampshire		X			Repeat for VLBW (<1500 g) & NICU at 2 weeks of age.
31 New Jersey		X			
32 New Mexico	X	X			
33 New York		X			3-5 days.
34 North Carolina		X			
35 North Dakota		X			
36 Ohio				X	
37 Oklahoma		X			3-5 days of age.
38 Oregon	X				
39 Pennsylvania		X			When initial test is an inconclusive range we recommend a repeat filter paper.
40 Rhode Island		X			
41 South Carolina		X			Recommended if specimen collected < 24 hours of age.
42 South Dakota		X			If sample taken < 24 hours.
43 Tennessee		X			
44 Texas	X				Required for all infants.
45 Utah	X				
46 Vermont		X			
47 Virginia		X			When antibiotics or transfusion are indicated or when interfering substances limit interpretation of results.
48 Washington					Second test recommended between 7 & 14 days.
49 West Virginia				X	
50 Wisconsin					Repeat recommended when initial collection is made prior to 24 hours of age.
51 Wyoming	NR				
52 Puerto Rico		X			< 48 hours
53 Virgin Islands					

Table 2.07: Laboratory Specimen Information

State/Territory	Total Number of Specimens Received	Number of Specimens Unacceptable for Analysis	% of Samples Deemed Unacceptable from Total Tests	Length of Time Blood Specimens Kept (room temperature unless noted)	Written policy for storage and Disposal of left-over Specimens (Yes/No)	Type of Computer Evaluation of Submissions	Type of Education Provided to Submitter
1 Alabama	189,383	14,543	7.68%	3 months at 2-8°C.	Yes	Neometrics Software	Quality assurance program.
2 Alaska	17,799	186	1.05%	Indefinitely	Yes-Lab keeps 1 yr & ships to state.	Matches births to specimens	Brochures, video's, presentations, and practitioners manual.
3 Arizona	141,967	1,941	1.37%	3 months	No	None	NBS professional staff provides in-service training.
4 Arkansas	37,260	180	0.48%	1 year	Yes	Queries compiled by lab rptng.	Inservice training for local health units and hospital staff.
5 California	527,297	3,392	0.64%	Indefinitely (-20°C)	Yes	Submitter compliance	
6 Colorado	123,749 c			3 months	Yes	None	NBS staff provides in-service training, videos, brochures.
7 Connecticut	87,663 b	151	0.17%	6 months	Yes	No	Written guidelines, In-services & site visits at birthing facilities.
8 Delaware	22,304	93	0.42%	4 mo. @ 2-8° range	Yes	Data shared via external modem.	Written procedures for handling specimens.
9 District of Columbia	15,412	124	0.80%	Indefinitely	Yes	Provided by lab.	Educational update in DC NBS Program made by Lab Director.
10 Florida	296,177	4,886	1.65%	- 5 years	No	Profile report sent to hospitals.	
11 Georgia	189,498	20,088	11%	6 weeks	No		Inservice training provided by program and lab staff.
12 Hawaii	17,612	16	0.09%	One year	Yes	Screening practice profiles	In-service training provided on specimen collection to submitters as necessary. Written material & video avail. upon request.
13 Idaho	34,260	237	0.69%	One year	Yes, OPHL policy NOT written into Idaho rules & reg.	Provided by OPHL.	Practitioners manual and parent brochures.
14 Illinois	188,164	648	0.34%	2 mo. , all specimens. 6 mo., positive specimens.	Yes	Monthly reports.	Brochures, videos, in-services, conferences. practitioners manual, workshops.
15 Indiana	119,421	2,676	2%	23 yrs. @ room temp.	No	Tracking of samples	NCCLS video on coll., S&S posters & onsite inservices.
16 Iowa	39,580	433	1.09%	4 weeks	Yes	None	Pamphlet written for parents, video for submitters.
17 Kansas	46,128	3,325	7.21%	30 days @ -10 - 20°C	Yes	Monthly evaluations	Phone consultations, site visits and conf. presentations.
18 Kentucky	78,449 c			6 mo. hypothyroid refrig.	Yes	Unsat. specimen rpt. generated.	Onsite inservice training, send out film & materials on req.
19 Louisiana	98,597	3,812	3.87%	2 weeks @ 4°C 1 month @ room temp.	Yes	None	Assist providers in developing NBS protocols and collection of satisfactory specimens.
20 Maine	13,341	81	0.61%	3 years	Yes	Specimen & date of receipt reports	In-service.
21 Maryland	152,077	7,487 a	4.92%	6 mo. desiccant @ -20°C 2½ yrs.	Yes	None	Continuing education, NCCLS video, & other literature.
22 Massachusetts	91,521	882	0.96%	1991 to present	Yes	Internal QC rpt. of specimens.	Inservice consultation of hosp, NCCLS video, website.
23 Michigan	144,489	2,182	1.51%	21.5 yrs	Yes	Quarterly reports.	Inservice training, and videos
24 Minnesota	68,410	1,460	2.13%	3 years	Yes	Internal QC rpt. of specimens.	Grand rounds, poster on collection, website, pamphlets.
25 Mississippi	44,075			3 months	Yes - Neometrics system	NBS lab test rejection reports are follow-up by MSDH each month.	Hospitals are in-serviced twice/yr, if repeat rate goes above 5% hosp. is in-serviced; each hosp. is evaluated every mo.
26 Missouri	97,217	1,678	1.73%	6 mos @ < -30°C	Yes	Neometrics MSDS	NCCLS video, phone consultation by lab personnel.

continued

Table 2.07: Laboratory Specimen Information (continued)

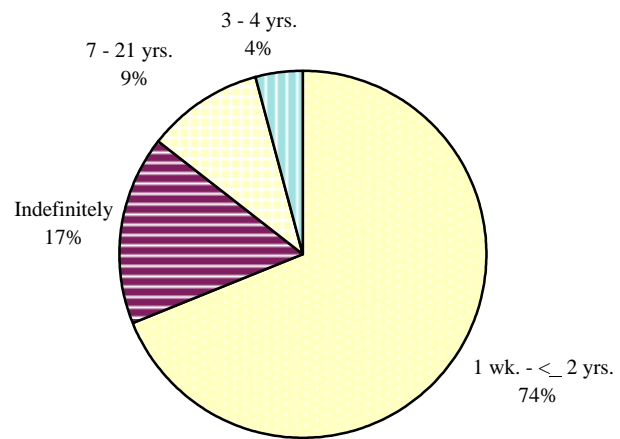
State/Territory	Total Number of Specimens Received	Number of Specimens Unacceptable for Analysis	% of Samples Deemed Unacceptable from Total Tests	Length of Time Blood Specimens Kept	Written policy for storage and Disposal of left-over Specimens (Yes/No)	Type of Computer Evaluation	Type of Education Provided
27 Montana	14,367			6-8 weeks 4°C	Yes	No computer evaluation.	Practitioners Manual, S&S posters, in-service on request.
28 Nebraska	24,863	33	0.13%		Yes, each lab has own policy		
29 Nevada	56,327	493	0.88%	1 year	Yes	Monthly rpts. sent from OR. PHL	Practice manual, brochures, on-site visit, CEU class etc.
30 New Hampshire	14,694	135	0.92%	7 years	Yes	Customized rpts as needed.	Hosp inservices, printed matter, Mass. Website.
31 New Jersey	122,975	185	0.15%	23 years	Yes	Submitter compliance.	On-site training, NCCLS video tape, practioners guidance manual.
32 New Mexico	49,993	654	1.31%	3 months	No	Computer profiles.	Pamphlets, practitioners manual, fact sheets, in-service.
33 New York	275,936	4,505	1.63%	6 mos. (20°C desiccant)	Yes	Annual statistics.	Video, posters, NBS guide, phone consultation, parent brochures.
34 North Carolina	135,349	1,494	1.10%	2 years	No	Unsat reports send to submitter	
35 North Dakota	9,300	136	1.46%	10 years	Yes	None	Pamphlet & video tape.
36 Ohio	197,587	3,311	1.68%	until tests completed	Yes		
37 Oklahoma	52,760	531	1.01%	4 weeks	Yes		NBS program rules/regulations mailed to all submitters.
38 Oregon	46,879	26	0.06%	1 year, room temp.	Yes	Screening Practice profiles sent monthly to hosp. admin. & mang.	Screening practice consultations, procedure & policy review provided by Educational coord., and NBS RN's.
39 Pennsylvania	148,597 c			5 years by State			Grand rounds presentation to the physician by contractor.
40 Rhode Island	13,577			23 years	Yes	Data by Hospital	QA. in service training.
41 South Carolina	53,620	1,466	2.73%	Indefinitely	Yes		Laboratory and follow-up training. On-site updates held at request of submitter.
42 South Dakota	11,998	32	0.27%	2 mo. @ 2.8°C in sealed bags	Yes		On site visits, written materials "Simple Spot Check"
43 Tennessee	84,110	6,318	7.51%	3 mo. 2-8°C; abn. indefinitely	Yes	Quarterly reports.	In-service edu, video & pamphlets to hospitals.
44 Texas	705,151			6 months	Yes	Monthly extraction of database	Practitioner's Guide, pamphlets, posters.
45 Utah	94,418	917	0.97%	3 months at 4°C	Yes	ND	No education provided by lab.
46 Vermont	6,366	73	1.15%	Indefinitely	No	Database w/HCP, hosp. lab, & nursery staff, hosp. QC every 6 mo.	Newsletters, in-service presentations for hosp. lab & nursery staffs, PH nurses by program coordinator.
47 Virginia	112,814	498	0.44%	nml = 6 mos; abn = 10 yrs	Yes	Submitter profiles.	Site visits, training seminars, literature prov, video.
48 Washington	147,624	1,138	0.77%	21 years	Yes	Quarterly & annual reports.	Educational on - site visits, instructions on blood coll.
49 West Virginia	31,990	2,247	7.02%		No	Monthly reports	NCCLS video.
50 Wisconsin	70,797	1,155	1.63%	1 year at 4 - 8 °C	No		Newsletter, NCCLS video, website, NBS manual.
51 Wyoming	8,892 c			3 months			
52 Puerto Rico	61,397	40	0.07%	6-12 months	Yes	Manual system	New hosp. initial training of 1-2 hours.
53 Virgin Islands	1,851	19	1.03%	12 months	Yes	with Program Director	Lecture, video, demonstrations to staff.

NBS = Newborn Screening, HCP = Health care providers, PH = Public Health

a = 1,364 were IMF specimens, 6,123 wer unsats. Unacceptable specimens are tested if results are normal, reported as unsatisfactory specimen; if results are abnormal, reported as abnormal but unsatisfactory specimen;

b = includes 1st, 2nd, and 3rd specimens; c = taken from number of specimens tested for PKU.

Figure 2.02: Specimen Storage Time - 1998



3. Hyperphenylalaninemia

3.0 Introduction

Screening for phenylketonuria has been a part of some U.S. screening programs since 1962. Over the years, as more has become known about the disorder, it has been generally accepted that the screening usually performed is for elevated levels of phenylalanine, or hyperphenylalaninemia. Of the hyperphenylalaninemias detected, the classical type is referred to as phenylketonuria (PKU). The tables in this section reflect information gathered from U.S. screening programs relative to detecting PKU and other hyperphenylalaninemias.

3.1 Definitions for Classical Phenylketonuria

Programs were asked to give the definition used by their program to define classical phenylketonuria. There is still not 100% agreement among programs as to the definition, based on serum laboratory testing. The vast majority of programs use a serum phenylalanine value of >20 mg/dL as definitive, but a few programs use lower values and others prefer to leave the diagnosis to individual physicians treating the disorder. Even in cases where states have reported serum values for the definition, they still rely on specialized treatment centers to make the final determination. The results of the survey of definitions used for classical PKU are given in Table 3.01.

3.2 Definitions for Clinically Significant Hyperphenylalaninemia Variants

In addition to detecting PKU, most programs utilize screening techniques that identify other hyperphenylalaninemia variants. Of these, some are considered to be clinically significant and require treatment and/or monitoring of serum phenylalanine levels. Programs were asked to list their definition of hyperphenylalaninemia variants that might fall into the category of clinically significant. Table 3.02 gives the results of this survey.

3.3 Definitions for Variant Hyperphenylalaninemias not Clinically Significant

So that programs could further differentiate between those cases of hyperphenylalaninemia that were clinically significant and those that were not, programs were asked to give their definition of 'not clinically significant.' Table 3.03 gives the responses to this question.

3.4 Laboratory Techniques

The bacterial inhibition assay developed by Bob Guthrie in the early 1960's was the assay of choice for almost all screening systems in their early days. While some programs used automated fluorometric procedures, only recently has there been more interest in developing better commercial procedures. The advantage of these methods is their ability to give more quantitative results than those of the semi-quantitative Guthrie procedure. In some cases they may also be more rapid. The disadvantage is usually their increased cost. Table 3.04 gives the responses of programs when asked about their current laboratory techniques.

3.5 Initial Screening Results

In order to ascertain the effectiveness of screening for hyperphenylalaninemias, programs were asked to report their findings on initial screening. Initial screening was defined as the first reportable test and programs were asked not to include duplicate screenings. By reviewing the number of newborns screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated. Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 3.05. Programs wishing to explain some of their responses included information included as footnotes to the table. Because some programs reported inaccurate or inappropriate data in some of their responses, totals were not given in some columns of the table. Care should be taken in using these data, since not all programs reported data.

3.6 Second Screen Results

In addition to initial screening results, most programs also report second screening results on at least some of their patients, and many even reported additional screening results beyond the second. In an effort to

evaluate the productivity of additional screening tests beyond the initial screen, programs were asked to report outcome of additional screening. The data were requested in three categories.

Programs that require or receive second testing on newborns regardless of the results of the first screening test either by law or, by rule, or by strong inference, were asked to report these data as 'second screens.' In Table 3.06, these data are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary'). Programs who requested an additional screen because of a certain result on the first screen that acted as impetus for another screen, or because of an unsatisfactory first screen (usually due to sample condition or early collection), were asked to report these data as 'repeat screens.' Care was taken to emphasize the point that 'repeat' specimens included those collected for purposes of clarifying an initial screening result.

For each case, the data requested included the total newborns tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. Once again, the reader is cautioned about using the data without regard to notes of explanation.

3.7 Data Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of classical phenylketonuria, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain and report Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 3.07. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

3.8 Clinically Significant Variants by Sex and Race/Ethnicity

Programs were also asked to report all clinically significant variants as noted in Section 3.7. These data are reported in Table 3.08.

3.9 Hyperphenylalaninemia Variants Not Clinically

Significant by Sex and Race/Ethnicity

Programs were asked to report totals of variants of hyperphenylalaninemia not considered to be clinically significant divided by sex and race/ethnicity. These data are reported in Table 3.09.

3.10 Time to Treatment

In order to look at overall program efficiency, programs were asked to report the number of days from birth until treatment of classical PKU. The definition of treatment used in the questionnaire was "initiation of dietary or drug regimen" and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 3.10.

3.11 Historic Data

In order to document the value of screening for hyperphenylalaninemia, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of various types of cases detected over the years. These data are given in Table 3.11

Since many programs have historically not maintained these data, the reader is cautioned about using them out of context.

3.12 Total Cases Detected

Table 3.12 gives a summation of the data contained in Tables 3.05 and 3.06 along with the number of births in each state or territory as reported in Table 1.01. This table was included so that a tabulation of these data could be viewed without having to refer to several tables.

3.13 Summation of Testing

Table 3.13 is a table summing the significant testing data from this Chapter. It includes the total births from Table 1.01, the total newborns reported as being screened, and the testing data reported in Tables 3.05 and 3.06. Comparative percentages have been provided at the request of a number of readers, but in many cases these data are based on estimates or on data that appear to contain duplicate information thus causing total compliance percentages to exceed 100%. Data responders are requested to review these data and adjust their responses to future questionnaires so that more meaning can be associated with the data. In particular, the data in this table seem to point out the difficulty of many programs in separating total samples received from total births screened (i.e. many programs cannot differentiate between first tests received and others that might have been submitted as independent second tests or repeat screens).

Table 3.01: Definitions for Classical Phenylketonuria

State/Territory	Definition for Classical Phenylketonuria
1 Alabama	Phenylalaninemia > 20 mg%.
2 Alaska	Phe > 20 mg/dL on regular diet.
3 Arizona	Serum phe of ≥ 10 mg/dL.
4 Arkansas	Determined by metabolic specialist, phe level in 8-20 mg/dL range for which dietary treatment is prescribed.
5 California	Persistently elevated phe requiring dietary intervention.
6 Colorado	Phe > 20.
7 Connecticut	Classified by treatment center.
8 Delaware	Diagnosis and classification determined by medical consultant.
9 District of Columbia	Elevated phenylalanine level.
10 Florida	Phe ≥ 20 mg/dL (serum).
11 Georgia	Significant elevation of Phe level which can only be reduced & maintained @ treatment level by strict adherence to dietary restriction of Phe and usually added tyrosine.
12 Hawaii	Phe levels ≥ 20 mg/dL will be treated dietary restrictions. Diagnosis made by medical consultant.
13 Idaho	Phe level > 6 mg/dL - will be treated with dietary restriction.
14 Illinois	Diagnosis and classification determined by pediatric medical consultant.
15 Indiana	Phe ≥ 2.9 mg/dL. Diagnosis & classification made by medical consultant, supported with diagnostic testing.
16 Iowa	Phe level ≥ 20 mg/dL on amino acid analysis.
17 Kansas	Phe hydroxylase enzyme has no activity.
18 Kentucky	Treatment if levels persistently > 8 mg/dL.
19 Louisiana	Patient requiring a special low phe medical food to maintain adequate nutrition and achieve a phe level between 2mg/dL-5mg/dL.
20 Maine	Phe > 20 mg/dL. Confirmed by medical consultant.
21 Maryland	Phe ≥ 20 mg/dL.
22 Massachusetts	Diagnosis and classification determined by metabolic consultant.
23 Michigan	Phe > 20 mg/dL requires strict dietary management.
24 Minnesota	Phe ≥ 20 mg/dL, final diagnosis made by metabolic specialist.
25 Mississippi	Defined by genetic centers; in general phe ≥ 20 mg/dL.
26 Missouri	Diagnosis and classification determined by medical consultant.
27 Montana	Blood phe rises above 20 mg/dL on normal diet.
28 Nebraska	Phe ≥ 20 mg/dL; bioppterin profile normal; final diagnosis by metabolism specialist.
29 Nevada	> 6 mg/dL (infant at any age) - no other tests abnormal; 4 mg/dL (infant < 48 hrs old); 4 mg/dL (infant > 48 hrs old).
30 New Hampshire	Diagnosis and classification made by metabolic consultant.
31 New Jersey	Dx & classification confirmed by med. consultant; persistent highly elevated phe levels, confirmatory dx testing treated by specialist.
32 New Mexico	Persistently elevated phenylalanine levels > 10 mg/dL. Diagnosis and classification made by endocrinology consultant, supported by diagnostic tests.
33 New York	Diagnosis and classification made by treatment center.
34 North Carolina	Diagnosis and classification made by medical consultant.
35 North Dakota	Diagnosis and classification made by medical consultant and/or reference lab performing the diagnostic test.
36 Ohio	Diagnosis & classification made by medical consultant.
37 Oklahoma	Phe ≥ 15 mg/dL.
38 Oregon	Not appropriate, defined as "Clinically Significant Hyperphenylalaninemia."
39 Pennsylvania	Phenylalanine level > 20 mg/dL.
40 Rhode Island	Diagnosis and classification determined by medical consultant.
41 South Carolina	Determined by specialist.
42 South Dakota	Diagnosis and classification made by medical consultant.
43 Tennessee	Defined by Genetic Center in general phe > 20 mg/dL.
44 Texas	Untreated serum phenylalanine > 20 mg/dL.
45 Utah	Phenylalanine level > 20 mg/dL on a regular diet.
46 Vermont	Persistent phe elevation 10 mg/dL; diet manipulation required to maintain phe 10 mg/dL.
47 Virginia	Persistently elevated phenylalanine supported by confirmatory diagnostic testing.
48 Washington	Blood phenylalanine levels which rise above 10 mg/dL with normal protein intake.
49 West Virginia	Blood phenylalanine levels > 20 mg/dL on normal diet.
50 Wisconsin	Diagnosis and classification made by treatment center.
51 Wyoming	Phe >20 on formula, probable if Phe >17 on breast milk. Tolerance for dietary phe high, dx reconsidered, if necessary challenge at 1 yr.
52 Puerto Rico	Patients with deficiency of phenylalanine hydroxylase and untreated levels of phenylalanine more than 20 mg/dL.
53 Virgin Islands	Phe > 20 mg/dL.

Table 3.02: Definitions for Clinically Significant Variant Hyperphenylalaninemia

State/Territory	Definition for Clinically Significant Variant Hyperphenylalaninemia
1 Alabama	Phenylalanine 6 mg%.
2 Alaska	Phe > 6 mg/dL % will be treated with dietary restriction.
3 Arizona	Serum Phenylalanine > 2 mg/dL.
4 Arkansas	Determined by metabolic specialist confirmed phe > 4 mg/dL but < 8-10 mg/dL.
5 California	Persistently elevated phe not requiring dietary intervention.
6 Colorado	Any phe > normal is significant & followed. Phe ≥ 10 on regular diet are treated w/restricted diet; < 10 on regular diet are monitored.
7 Connecticut	Classified by treatment center.
8 Delaware	Diagnosis and classification determined by medical consultant.
9 District of Columbia	Elevated phenylalanine level.
10 Florida	DA
11 Georgia	Moderate; some elevation of Phe which can only be kept within treatment range by moderate dietary intervention.
12 Hawaii	Phe levels ≥ 6mg/dL will be treated dietary restrictions. Diagnosis made by medical consultant.
13 Idaho	"Clin. Significant Hyperphenylalaninemia" - Phe > 4mg/dL. Diagnosis/classification made by med. consultant supported by diag. tests.
14 Illinois	Diagnosis and classification determined by pediatric medical consultant.
15 Indiana	Diagnosis & classification made by medical consultant.
16 Iowa	Persistent Phe levels ≥ 6 mg/dL.
17 Kansas	DA
18 Kentucky	Phe 20mg/dL %.
19 Louisiana	Untreated person with phe values ranging from 6-17 mg/dL without protein restricted intake.
20 Maine	Phe 4-6 mg/dL while on regular protein intake. Confirmed by medical consultant.
21 Maryland	Phe > 8 mg/dL but ≤ 20 mg/dL.
22 Massachusetts	Diagnosis and classification determined by metabolic consultant.
23 Michigan	Variable Hyperphe in infants with impaired bipterin metabolism due to specific bipterin enzyme def & not due to phe hydroxylase def.
24 Minnesota	Phe > 8 but < 20 mg/dL that remains at this level on retest, diagnosis made by medical specialist.
25 Mississippi	Defined by genetic centers; in general phe > 10-20mg/dL.
26 Missouri	Diagnosis and classification determined by medical consultant.
27 Montana	Excluding classical PKU, levels between 4-20 mg/dL.
28 Nebraska	NR
29 Nevada	Any elevations of PA.
30 New Hampshire	Diagnosis and classification made by metabolic consultant.
31 New Jersey	Dx & class. confirmed by med. consultant; persistent moderately elev. phe levels by confirmatory dx testing by specialist.
32 New Mexico	Phe ≥ 3 mg/dL but < 10 mg/dL, diagnosis & classification made by medical consultant, supported by diagnostic tests.
33 New York	Diagnosis and classification determined by treatment center.
34 North Carolina	Diagnosis and classification made by medical consultant.
35 North Dakota	Diagnosis and classification is determined by medical consultant.
36 Ohio	Diagnosis & classification made by medical consultant.
37 Oklahoma	Phe ≥ 6 and < 15 mg/dL, or physician diagnosis.
38 Oregon	Phe level > 6 mg/dL will be treated with dietary restrictions.
39 Pennsylvania	Phenylalanine level of 6 mg/dL to 19 mg/dL.
40 Rhode Island	Diagnosis an classification made by medical consultant.
41 South Carolina	Determined by specialist.
42 South Dakota	Diagnosis and classification made by medical consultant.
43 Tennessee	Defined by Genetic center in general phe > 4.0 - 10 mg/dL.
44 Texas	Untreated serum phenylalanine 10 - 19.9 mg/dL; maternal > 6 mg/dL.
45 Utah	Phenylalanine level > 10 mg/dL and < 20 mg/dL on a regular diet.
46 Vermont	Tetrahydrobiopterin deficiency associated with elevated blood phe levels (6-60 mg/dL).
47 Virginia	Phe < 6 mg/dL on full protein diet; no treatment male or female.
48 Washington	Blood phenylalanine levels which remain between 6 - 10 mg/dL with normal protein intake.
49 West Virginia	Blood phenylalanine levels of 10 - 20 mg/dL.
50 Wisconsin	Diagnosis and classification made by treatment center.
51 Wyoming	Phe level > normal is signif. & is followed; levels >12 on reg. diet treated w/restricted diet; levels between 10-12 on reg. diet options for treatment are discussed with family; <10 on reg. diet are monitored.
52 Puerto Rico	Phe blood levels > 10 - < 20mg/dL in untreated patients. Patients with Phe levels between 4-10 mg/dL without treatment are considered hyperphenylalaninemia. They do not require treatment, although we do restrict protein intake.
53 Virgin Islands	Phe > 8 mg/dL but < 20 mg/dL.

Table 3.03: Definitions for Variant Hyperphenylalaninemia - Not Clinically Significant

State/Territory	Definition for Variant Hyperphenylalaninemia - Not Clinically Significant
1 Alabama	N/R
2 Alaska	Phe < 6 mg/dL %.
3 Arizona	All variants are considered to be clinically significant.
4 Arkansas	Determined by metabolic specialist, phe level consistently less than 8 mg/dL for which no treatment is prescribed.
5 California	DA
6 Colorado	DA
7 Connecticut	Classified by treatment center.
8 Delaware	Diagnosis and classification determined by medical consultant.
9 District of Columbia	Elevated phenylalanine level.
10 Florida	Phe > 10 mg/dL or < 20 mg/dL.
11 Georgia	Slightly elevated Phe level which does not require restricted diet to maintain within treatment range.
12 Hawaii	Phe levels consistently < 6 mg/dL. Diagnosis made by medical consultant.
13 Idaho	Phe level consistently under 6 mg/dL%.
14 Illinois	Diagnosis and classification determined by pediatric medical consultant.
15 Indiana	Diagnosis & classification made by medical consultant.
16 Iowa	Phe > 4 mg/dL and < 6 mg/dL.
17 Kansas	DA
18 Kentucky	DA
19 Louisiana	Untreated person with phe values ranging from 3-5 mg/dL.
20 Maine	Phe 6-20 mg/dL while on regular protein intake. Confirmed by medical consultant.
21 Maryland	Phe > 4 mg/dL but ≤ 8 mg/dL.
22 Massachusetts	Diagnosis and classification determined by metabolic consultant.
23 Michigan	Persistent Phe levels never exceeding 10 mg/dL without diet restriction - clinical significance in adult years cannot be determined.
24 Minnesota	Phe > 4 mg/dL but < 8 mg/dL that remains at this level on retest, final diagnosis made by medical specialist.
25 Mississippi	Defined by genetic centers; in general phe > 4-10mg/dL.
26 Missouri	Diagnosis and classification determined by medical consultant.
27 Montana	Phe concentration of 3-4 mg/dL.
28 Nebraska	Phe ≥ 2 mg/dL but < 4 mg/dL; biopterin profile normal.
29 Nevada	NR
30 New Hampshire	Diagnosis and classification made by metabolic consultant.
31 New Jersey	Dx & class. confirmed by med. consultant; persistent mildly elev. phe levels confirmatory dx testing , require treatment pregnancies only.
32 New Mexico	Diagnosis and classification made by confirmatory labs and endocrinology consultant.
33 New York	Diagnosis and classification determined by treatment center.
34 North Carolina	Diagnosis and classification made by medical consultant.
35 North Dakota	Phe > 4 mg/dL and < 10 mg/dL.
36 Ohio	Diagnosis & classification made by medical consultant.
37 Oklahoma	Phe ≥ 4 and < 6 mg/dL, or physician diagnosis.
38 Oregon	Phe level consistently under 6 mg/dL.
39 Pennsylvania	Phenylalanine level of 5 mg/dL.
40 Rhode Island	Diagnosis and classification made by medical consultant.
41 South Carolina	Determined by specialist.
42 South Dakota	Diagnosis and classification made by medical consultant.
43 Tennessee	Defined by Genetic center in general phe > 10-20 mg/dL.
44 Texas	Untreated serum phenylalanine 4 - 9.9 mg/dL.
45 Utah	Phenylalanine level > 2.1 mg/dL and < 10 mg/dL on a regular diet.
46 Vermont	Blood phe levels persistently 4-10 mg/dL without diet manipulation.
47 Virginia	Phe > 6 mg/dL on full protein diet, no associated urinary ketones. Females follow into reproductive adulthood.
48 Washington	Blood phenylalanine levels which remain between 3 & 6 mg/dL with normal protein intake.
49 West Virginia	Blood phenylalanine levels > 4 mg/dL but < 10 mg/dL with protein intake.
50 Wisconsin	Diagnosis and classification made by treatment center.
51 Wyoming	Phe level > normal is signif. & is followed; levels >12 on reg. diet treated w/restricted diet; levels between 10-12 on reg. diet options for treatment are discussed with family; <10 on reg. diet are monitored.
52 Puerto Rico	Phenylalanine blood levels of 4 - 10 mg/dL.
53 Virgin Islands	Phe > 4 mg/dL but < 8 mg/dL.

Table 3.04: Hyperphenylalaninemia - Laboratory Testing

State/Territory	Microbiological Inhibition Assay (Guthrie)	Fluorometric	Other	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Immediate Serum Follow-up</i>
1 Alabama	X			> 4 mg/dL	DA
2 Alaska	X			≥ 4 mg/dL	
3 Arizona	X			Jan-Jun ≥ 3 mg/dL; July-Dec 2-3 inconclusive ≥ 3 abn.	First screen ≥ 6 mg/dL or second abnormal
4 Arkansas		X		4 - 8 mg/dL	≥ 8 mg/dL
5 California		X		Phe/tyr ≥ 1.5 and phe ≥ 150 μM	NC
6 Colorado	X			≥ 4 mg/dL	Initial ≥ 6 mg/dL; recall ≥ 4 mg/dL
7 Connecticut		X		> 2 mg/dL but ≤ 4.0 mg/dL	≥ 6.0 mg/dL
8 Delaware			Colorimeter	Refer all infants with > 4 mg/dL	Refer all infants with > 4 mg/dL
9 District of Columbia			MS/MS		≥ 2 mg/dL
10 Florida		X		2.1 - 2.9 repeat done on same specimen.	≥ 2.5 mg/dL
11 Georgia	X			≥ 4, < 6	≥ 6
12 Hawaii	X			≥ 4 mg/dL	6 mg/dL
13 Idaho	X			4.0 mg/dL	> 6 mg/dL
14 Illinois		X		4.0 mg/dL	
15 Indiana		X		≥ 2.9 mg/dL	≥ 6.0 mg/dL
16 Iowa		X		3.1 mg/dL	> 10 mg/dL
17 Kansas		X		2.1 - 2.99 mg/dL	≥ 3 mg/dL
18 Kentucky	X				
19 Louisiana		X		2.5 - 2.9 mg/dL	3 mg/dL
20 Maine			MS/MS	> 3 mg/dL	> 5 mg/dL
21 Maryland	X		HPLC	Phe 2 & 4 mg/dL; Tyrosine < 12 mg/dL.	Phe 4 mg/dL
22 Massachusetts			MS/MS	> 2.3 mg/dL	All elevations ≥ 3 mg/dL reported by phone
23 Michigan		X		2.0 - 2.9 mg/dL < 24 hrs; 2.1-2.9 < 24 hrs	3.0 mg/dL
24 Minnesota	X			≥ 4 mg/dL	≥ 2 mg/dL
25 Mississippi		X		4.0-5.0 mg/dL	5.0 mg/dL
26 Missouri		X		3 mg/dL	5 mg/dL
27 Montana		X		> 3.0 mg/dL	≥ 25 mg/dL
28 Nebraska c		X			≥ 3.4
29 Nevada	X			≥ 4 mg/dL > 48 hours old	> 4 mg/dL < 48 hrs; > 6mg/dL any age.
30 New Hampshire			MS/MS	> 3 mg/dL	All elevations reported by phone.
31 New Jersey		X		≥ 2.1-5.9 mg/dL	> 6 mg/dL
32 New Mexico	X	X		≥ 3 μg/dL	significant increase level referred to physician for follow-up.
33 New York	X		X d	≥ 3 mg/dL and < 6 mg/dL	6 mg/dL
34 North Carolina			MS/MS	157 μM	DA
35 North Dakota		X e		≥ 3.1 mg/dL	≥ 10 mg/dL
36 Ohio		X		FIA 2.1 - 2.7	FIA ≥ 2.7
37 Oklahoma			Enzyme assay	≥ 3.5 mg/dL	> 5.0 mg/dL
38 Oregon	X			≥ 4.0% mg/dL	
39 Pennsylvania			MS/MS		
40 Rhode Island			MS/MS	≥ 2.3 mg/dL	≥ 3 mg/dL
41 South Carolina f		X		≥ 4.0 mg/dL	DA
42 South Dakota				4 - 10 mg/dL	> 10 mg/dL
43 Tennessee		X		> 4.0 mg/dL	> 5.0 mg/dL
44 Texas	X a	X b		All not normal are serum tested	≥ 4.0 mg/dL
45 Utah		X		≥ 2.1 mg/dL	2 abnormal screens
46 Vermont			MS/MS	3 to < 6 mg/dL	≥ 6 mg/dL
47 Virginia	X		HPLC	≥ 4 mg/dL	≥ 10 mg/dL
48 Washington	X		HPLC	≥ 4 mg/dL; ≥ 3 mg/dL if ≤ 24 hours	≥ 8 mg/dL
49 West Virginia		X		≥ 4 mg/dL	
50 Wisconsin		X		≥ 2.2 mg/dL	2nd filter paper abnormal
51 Wyoming	X			≥ 4 mg/dL	Initial ≥ 6 mg/dL; recall ≥ 4 mg/dL
52 Puerto Rico	X		HPLC	4-10 mg/dL phenylalanine	10 mg/dL phenylalanine
53 Virgin Islands	X				

a = initial screen; b = retest on abnormal; c = Delfia used by 2 laboratories, other laboratory uses flurometer; d = thin layer chromatography is used as a confirmatory test for phenylalanine levels > 6 mg/dL; e = Isolab; f = all results > 4.0 mg/dL on previously undiagnosed infants are called to the physician of record.

Table 3.05: Initial Screening Results - Hyperphenylalaninemia

State/Territory	Number of Newborns Screened	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up	Number of Newborns Confirmed with Classical PKU	Number of Newborns Confirmed w/Clin. Sig. Variant	Number of Newborns Confirmed w/ Variant (Not Clin. Sig.)
1 Alabama	62,173	7	0	4	0	0
2 Alaska	9,821	1	0	0	0	0
3 Arizona	81,956	31	1	4	1	NA
4 Arkansas	36,446	5	0	0	0	0
5 California	527,297	188	0	18	21	0
6 Colorado	63,219	42	0	4	2	DA
7 Connecticut	43,723	38	0	2	0	6
8 Delaware	11,714	1	NA	3	0	1
9 District of Columbia	15,125	4	1 c	0	NA	NA
10 Florida	204,030	109	0	15	4	0
11 Georgia	189,498		0	5	2	0
12 Hawaii a	17,612	1	0	0	0	0
13 Idaho	20,254	5	0	3	0	0
14 Illinois b	188,164	234	1	10	2	7
15 Indiana	87,639	81	2	10	5	3
16 Iowa	38,141	16	1 h	4	0	2
17 Kansas	39,031		0	1	0	DA
18 Kentucky	54,515	9	N/A	4		
19 Louisiana	67,843	24	0	2	1	0
20 Maine	13,341	3	0	1	0	0
21 Maryland	72,390	79	0	2	1	1
22 Massachusetts	82,703	44	NA	1		5
23 Michigan	134,022	197	0	5	3 g	2
24 Minnesota	68,402	13	0	3	2	0
25 Mississippi	44,075	29	0	1	0	0
26 Missouri	78,640	31	0	7	1	0
27 Montana	10,825	1	0	0	0	0
28 Nebraska	24,863	6	1 h	1	2	2
29 Nevada	30,659	12	0	2	0	0
30 New Hampshire	13,879	8	0	0	0	0
31 New Jersey	112,241	166	0	5	0	5
32 New Mexico	25,865	31	0	0	0	
33 New York	258,449	205	1	14	11	9
34 North Carolina f	120,750	NC	ND	2	6	ND
35 North Dakota	8,806	3	0	0	0	0
36 Ohio	160,566	138	0	9	1	2
37 Oklahoma b	52,760	24	0	2	0	1
38 Oregon	46,879	15	0	4	1	0
39 Pennsylvania	148,597	44		10	5	3
40 Rhode Island	13,150	1	0	0	1	0
41 South Carolina	52,176	26	0	3	0	2
42 South Dakota	10,760	64	0	0	1	0
43 Tennessee	79,539	58	0	2	1	0
44 Texas	355,100	NC	NC	11	4	1
45 Utah	47,423	36	1	0	0	2
46 Vermont	6,040	3	0	1	0	1
47 Virginia	99,410	15	0	1	0	0
48 Washington	76,886	12	0	6	0	3
49 West Virginia	21,321	2	0	0	0	0
50 Wisconsin	66,614	38	0	7	0	2
51 Wyoming	5,480	1	0	0	0	0
52 Puerto Rico	57,155	634	101	3	0	0
53 Virgin Islands	1,851	19	0	0	0	0
TOTAL	e	e	e	192 d	78 d	60 d

a = Bioppterin testing done only on confirmed cases of clinically significant hyperphe and variant hyperphe; b = includes initial and repeat filter paper specimens submitted; c = lab lost to follow-up - DC MCH has more info; d = total given does not include all states; e = totals not given - too many programs reported inaccurate or inappropriate data; f = number of specimens rather than number of newborns collected; g = 3 mild not hyperphe; h = expired.

Table 3.06: Second Screens for Hyperphenylalaninemia

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with NOT NORMAL Test Results				Number of Newborns with NOT NORMAL Test Results Lost to Follow-up				Number of Newborns Confirmed with Classical PKU				Number of Newborns Confirmed w/Clin. Sig. Variant				Number of Newborns Confirmed w/ Variant (Not Clin. Sig.)			
	R	D	P		Total	R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total			
1 Alabama		61,927		61,927																				
2 Alaska		7,759	215	7,974																				
3 Arizona		57,352		57,352	13			13	0	0	0		0	0	0		1	0	1		0	0	0	
4 Arkansas			558	558			0	0			0	0			0	0			0	0			0	0
5 California			3,372	3,372				0			0	0			0	0			0	0			0	0
6 Colorado	60,530			60,530	5		N/C				N/C				N/C				N/C				N/C	
7 Connecticut		43,942		43,942	12			12	0		0		0		0		0		0		0		0	0
8 Delaware	10,692			10,692			0	N/A			N/A			0	0	0	0		0	0	0		0	0
9 District of Columbia			287	287			1	1			1	1	a		0	0			0	0			0	0
10 Florida				0				N/A				N/A			N/A				N/A				N/A	
11 Georgia		N/D	N/D	0				0			0				0				0				0	
12 Hawaii		DA	187	187			0	0			0	0			0	0			0	0			0	0
13 Idaho		13,804	199	14,003	3	2		5					1		1		0		0					
14 Illinois		N/A	N/C	0																				
15 Indiana		DA	31,782	31,782			64	64			1	1			0			0	0			0	0	
16 Iowa		DA	1,439	1,439		DA		0		DA	0	0		DA	0	0		DA	0	0		DA	0	0
17 Kansas				0	0			0				0							0				0	
18 Kentucky	NR																							
19 Louisiana				0		0	0	0		0	0	0		0	0	0		0	0	0		0	0	0
20 Maine																								
21 Maryland	61,152			61,152	4			4					0		1			1		2			2	
22 Massachusetts			8,818	8,818			42	42			0			0				0					0	
23 Michigan				0				0			0	0			0			1	0	1	e	6		6
24 Minnesota			619	619			13	13																
25 Mississippi		0		0	0			0	0	0			0	0			0		0			0		0
26 Missouri		13,295	5,282	18,577				0																

continued

Table 3.06: Second Screens for Hyperphenylalaninemia (continued)

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen R	Newborns Receiving Discretionary Second Screens D	Newborns Receiving Repeat Second Screens P	Total 2nd Screens c	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed with Classical PKU				Number of Newborns Confirmed w/Clin. Sig. Variant				Number of Newborns Confirmed w/ Variant (Not Clin. Sig.)						
					R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total			
27 Montana		3,550		3,550																							
28 Nebraska		DA	N/C			DA	N/C																				
0 Nevada	25,668			25,668	1			1																			
30 New Hampshire		815		815																							
31 New Jersey		DA	10,716	10,716		DA	37	37		DA	0	0		DA	0	0		DA	0	0		DA	0	0			
32 New Mexico	23,851			23,851	5		0	5																			
33 New York		DA		0		DA	N/C			DA	N/C			DA	N/C			DA	N/C			DA	N/C				
34 North Carolina		N/C	N/C		0			0		N/D	N/D			DA	DA			DA	DA			DA	DA				
35 North Dakota		DA	494	494		DA		0		DA	0	0		DA	0	0		DA	0	0		DA	0	0			
36 Ohio			37,021	37,021									0	0	0			N/C	N/C			N/C	N/C				
37 Oklahoma		N/C	N/C			N/C	N/C			N/C	N/C			N/C	N/C			N/C	N/C			N/C	N/C				
38 Oregon	42,423		861	43,284	4		1	5	0		0	0	0		0	0					0		0	0			
39 Pennsylvania																											
40 Rhode Island			427	427			1	1																			
41 South Carolina			3,657	3,657				0		0	0	0		0	0	0		0	0	0		0	0	0			
42 South Dakota		1,583	102	1,685		8	2	10																			
43 Tennessee				0																							
44 Texas	348,132		1,919	350,051				0				0								1			1				
45 Utah	46,460		387	46,847	10			10			0	0	0		0	0	0		0	0	0	2	0	2			
46 Vermont			326	326			1	1					0										0				
47 Virginia			12,644	12,644			5	5			0				0			0	0			0	0				
48 Washington		69,588	1,150	70,738		6	NR	6		0	NR	0		0	NR	0		0	NR	0		0	NR	0			
49 West Virginia			N/A	0		1		1		0		0					0	0	0			0	0	0			
50 Wisconsin		1,196	1,142	2,338		1	10	11		0	0	0		0	0	0		0	0	0		0	0	0			
51 Wyoming			3,413	3,413	DA	N/C	N/C		DA	N/C	N/C		DA	N/C	N/C		DA	N/C	N/C		DA	N/C	N/C				
52 Puerto Rico			533	533			3	3																			
53 Virgin Islands		40	29	69																							
Total	d	618,908	274,851	127,579	c			c				c	0	1	0	1	b	1	2	0	3	b	5	6	0	11	b

a = lab lost to follow-up-DC MCH has more info; b = total given does not include all states; c = totals not given - too many programs reported inaccurate or inappropriate data; d = totals given reflect program responses and should be viewed as estimates only, given the number not reporting and the number of caveats listed; e = 1 mild.

Table 3.07: Cases of Classical Phenylketonuria

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Alabama	1	3													4		
2 Alaska																		
3 Arizona	1	2							1				2	2	4	1		1
4 Arkansas																		
5 California	10	1									1	10	2	18 ³	1	5	6	
6 Colorado a											1	3	1	3	4			
7 Connecticut	1	1											1	1	2			
8 Delaware	3												3	0	3			
9 District of Columbia																		
10 Florida	4	6								3	2	7	8	15				
11 Georgia	3	2											3	2	5			
12 Hawaii																		
13 Idaho	2	2											2	2	4			
14 Illinois	3	1							1		2	1	6	2	10 ³	1	1	2
15 Indiana	4	5											4	5	10 ³		1	1
16 Iowa	4												4	0	4			
17 Kansas	1												1	0	1			
18 Kentucky											1	3	1	3	4			
19 Louisiana		2											0	2	2			
20 Maine		1											0	1	1			
21 Maryland		1	1										1	1	2			
22 Massachusetts											1	0	1	1				
23 Michigan	2	3											2	3	5			
24 Minnesota	1	2											1	2	3			
25 Mississippi	1												1	0	1			
26 Missouri	4	3											4	3	7			
27 Montana																		
28 Nebraska	1												1	0	1			
29 Nevada		2											0	2	2			
30 New Hampshire																		
31 New Jersey	2	2		1									2	3	5			
32 New Mexico																		
33 New York	6	5						1	1				7	6	14 ³	1	1	1
34 North Carolina		1				1							0	2	2			
35 North Dakota																		
36 Ohio	3	2									1	3	4	5	9			
37 Oklahoma	2												2	0	2			
38 Oregon	3	1											3	1	4			
39 Pennsylvania	5	4				1							6	4	10			
40 Rhode Island																		
41 South Carolina	2	1											2	1	3			
42 South Dakota																		
43 Tennessee		2											0	2	2			
44 Texas	5	4											5	4	11 ³	2	2	2
45 Utah																		
46 Vermont		1											0	1	1			
47 Virginia		1											0	1	1			
48 Washington	3	2							1				4	2	6			
49 West Virginia																		
50 Wisconsin	5	2											5	2	7			
51 Wyoming																		
52 Puerto Rico									2	1			2	1	3	2	1	3
53 Virgin Islands																		
TOTAL	82	65	1	1	1	1	0	1	6	1	8	14	98	83	193	5	11	16

¹ did not report a breakdown of Race/Ethnicity.

² total reported is from Table 3.05 and Table 3.06; therefore total does not equal or may differ from data given in Table 3.07 and denotes a problem in reliability.

³ Hispanic counted as a Race and is included in total.

a = race/ethnicity data not collected.

Table 3.08: Cases of Variant Hyperphenylalaninemia (Clinically Significant)

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Alabama																	
2 Alaska																		
3 Arizona		1			1							1	1	2		1	1	
4 Arkansas																		
5 California	6	4		1			1				1	1	7	7	21 ¹	4	3	7
6 Colorado a											2		2	0	2			
7 Connecticut																		
8 Delaware																		
9 District of Columbia																		
10 Florida	1	1									1	1	2	2	4			
11 Georgia	2												2	0	2			
12 Hawaii																		
13 Idaho																		
14 Illinois		1											0	1	2 ³	1		1
15 Indiana	3	1			1								4	1	5			
16 Iowa																		
17 Kansas																		
18 Kentucky																		
19 Louisiana		1											0	1	1			
20 Maine																		
21 Maryland	2												2	0	2			
22 Massachusetts																		
23 Michigan b	3	1											3	1	4			
24 Minnesota	1	1											1	1	2			
25 Mississippi																		
26 Missouri		1											0	1	1			
27 Montana																		
28 Nebraska															2 ¹			
29 Nevada																		
30 New Hampshire																		
31 New Jersey																		
32 New Mexico																		
33 New York	5	5											5	5	11 ³	1		1
34 North Carolina	3	2	1										4	2	6			
35 North Dakota																		
36 Ohio											1		1	0	1			
37 Oklahoma																		
38 Oregon	1												1	0	1			
39 Pennsylvania	2	3											2	3	5			
40 Rhode Island		1											0	1	1			
41 South Carolina																		
42 South Dakota	1												1	0	1			
43 Tennessee	1												1	0	1			
44 Texas	2	2											2	2	4			
45 Utah																		
46 Vermont																		
47 Virginia																		
48 Washington																		
49 West Virginia																		
50 Wisconsin																		
51 Wyoming																		
52 Puerto Rico																		
53 Virgin Islands																		
TOTAL	33	25	1	1	2	0	0	1	0	0	5	2	41	29	81	6	4	10

¹ did not report a breakdown of Race/Ethnicity.

² total reported is from Table 3.05 and Table 3.06; therefore total does not equal or may differ from data given in Table 3.08 and denotes a problem in reliability.

³ Hispanic counted as a Race and is included in total.

a = race/ethnicity data not collected; **b** = we use variant only for BH4 deficient infants.

Table 3.09: Cases of Variant Hyperphenylalaninemia (Not Clinically Significant)

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Alabama																	
2 Alaska																		
3 Arizona																		
4 Arkansas																		
5 California																		
6 Colorado																		
7 Connecticut	2	4											2	4	6		1	1
8 Delaware	1												1	0	1			
9 District of Columbia																		
10 Florida																		
11 Georgia																		
12 Hawaii																		
13 Idaho																		
14 Illinois	3	2											3	2	7 ¹	1	1	2
15 Indiana	1	1											1	1	3 ³		1	1
16 Iowa	1	1											1	1	2			
17 Kansas																		
18 Kentucky																		
19 Louisiana																		
20 Maine																		
21 Maryland	2								1				3	0	3			
22 Massachusetts															5 ¹			
23 Michigan	6	1	1										7	1	8			
24 Minnesota																		
25 Mississippi																		
26 Missouri																		
27 Montana																		
28 Nebraska															2 ¹			
29 Nevada																		
30 New Hampshire																		
31 New Jersey	2	2			1								3	2	5			
32 New Mexico																		
33 New York	4	3	1										5	3	9 ³	1		1
34 North Carolina															ND			
35 North Dakota																		
36 Ohio															2 ¹			
37 Oklahoma							1						1	0	1			
38 Oregon																		
39 Pennsylvania	1	2											1	2	3		1	1
40 Rhode Island																		
41 South Carolina	1	1											1	1	2			
42 South Dakota																		
43 Tennessee																		
44 Texas	1												1	0	2 ³	1		1
45 Utah												4	0	4	4			
46 Vermont		1											0	1	1			
47 Virginia																		
48 Washington	3												3	0	3	2		2
49 West Virginia																		
50 Wisconsin	1	1											1	1	2			
51 Wyoming																		
52 Puerto Rico																		
53 Virgin Islands																		
TOTAL	29	19	2	0	1	0	1	0	1	0	0	4	34	23	71	5	4	9

¹did not report a breakdown of Race/Ethnicity.

²total reported is from Table 3.05 and Table 3.06; therefore total does not equal or may differ from data given in Table 3.09 and denotes a problem in reliability.

³Hispanic counted as a Race and is included in total.

Table 3.10: Days from Birth Until Treatment Initiated for Classical Phenylketonuria

Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	TOTAL
1 Alabama																4 ¹
2 Alaska																0
3 Arizona			1	1		1								1		4
4 Arkansas																0
5 California			2	2	2	2	1	4	1	1			1	1	1	18
6 Colorado									3					1		4
7 Connecticut					1	1										2
8 Delaware	1		1	1												3
9 District of Columbia																0
10 Florida	1	1				2	1	2				2	3	3		15
11 Georgia	2						1	1					1			5
12 Hawaii																0
13 Idaho					1	1	1							1		4
14 Illinois			1	1					1		2		2	2	1	10
15 Indiana		1		1	1	3	1	1				1		1		10
16 Iowa			1						1		1		1			4
17 Kansas								1								1
18 Kentucky															4	4
19 Louisiana							1				1					2
20 Maine				1												1
21 Maryland					1		1									2
22 Massachusetts															1	1
23 Michigan		1							4							5
24 Minnesota					1	1				1						3
25 Mississippi														1		1
26 Missouri			1		1			2					3			7
27 Montana																0
28 Nebraska				1												1
29 Nevada															2	2
30 New Hampshire																0
31 New Jersey															5	5
32 New Mexico																0
33 New York	1			4	1	3	1	3		1						14
34 North Carolina															2	2
35 North Dakota																0
36 Ohio															9	9
37 Oklahoma													1	1		2
38 Oregon			1	1					1				1			4
39 Pennsylvania			1	3	2		1						2	1		10
40 Rhode Island																0
41 South Carolina		1		1					1							3
42 South Dakota																0
43 Tennessee															2	2
44 Texas											2	3	6			11
45 Utah																0
46 Vermont								1								1
47 Virginia	1															1
48 Washington				1	1	1				1			2 ^a			6
49 West Virginia																0
50 Wisconsin			2	1	3	1										7
51 Wyoming																0
52 Puerto Rico													3			3
53 Virgin Islands																0

¹did not report a breakdown of Days from Birth.

²total reported is from Table 3.05 and Table 3.06; therefore total does not equal or may differ from data given in Table 3.10 and denotes a problem in reliability.

a = one born out-of-hospital & NBS test delayed 13 days, one baby was referred to OR treatment was delayed there.

Table 3.11: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Classical Phenylketonuria			Variant Hyperphenylalaninemia Clinically Significant			Variant Hyperphenylalaninemia Not Clinically Significant		
	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases
1 Alabama	1963	37	91						
2 Alaska	1987	13	12	1987	13	3	1987	13	2
3 Arizona	1994	6	23	1994	6	16			
4 Arkansas	1973	27	56	1973	27	17	DA		DA
5 California	1980	20	394	1980	20	365	DA		DA
6 Colorado	Mar-79	20 3/4	51	Mar-79	20 3/4	61			
7 Connecticut	1996	4	149	1996	4	4	1985	15	40
8 Delaware	1993	7	7	1993	7	2	1993	7	3
9 District of Columbia	1996	4	1	1996	4	0	1996	4	0
10 Florida	1965	35	176	1965	35	81	DA		DA
11 Georgia	Sep-78	21 1/4	88	Sep-78	21 1/4	53			
12 Hawaii	1986	14	9	1986	14	3	1986	14	3
13 Idaho	1962	38	69						
14 Illinois	1965	35	409	1965	35	153	1965	35	7
15 Indiana	Jul-85	15 1/2	67	Jul-85	15 1/2	28	2000	0	3
16 Iowa	N/C		N/C	N/C		N/C	N/C		N/C
17 Kansas	1965	35	185	1993	7	2	1998	2	1
18 Kentucky NR									
19 Louisiana	1980	20	78	N/C		N/C	N/C		N/C
20 Maine	1976	24	32	1976	24	11	N/D		N/D
21 Maryland	1965	35	123	1993	7	54	1993	7	22
22 Massachusetts	1962	38	255	N/A		N/A	N/A		N/A
23 Michigan	1965	35	171	1965	35	91	1965	35	221
24 Minnesota NR									
25 Mississippi	1982	18	29	1982	18	34	1982	18	30
26 Missouri NR									
27 Montana	1982	18	16	1982	18	3	N/C		N/C
28 Nebraska	1989	11	14 a	N/C		N/C	N/C		N/C
29 Nevada NR									
30 New Hampshire	1983	17	13	1983	17	3	N/C		N/C
31 New Jersey NR									
32 New Mexico	1981	19	16	N/A		N/A	N/A		N/A
33 New York	1965	35	525	1996	4	68	1998	2	20
34 North Carolina NR									
35 North Dakota	1964	36	31						
36 Ohio NR									
37 Oklahoma	1991	9	19	NC		N/C	N/C		N/C
38 Oregon NR									
39 Pennsylvania NR									
40 Rhode Island	1976	24	20	N/A	N/A	N/A	N/C	N/C	N/C
41 South Carolina	1962	38	96 a	1962	38	4 a	1962	38	4 a
42 South Dakota NR									
43 Tennessee				N/C		N/C	N/C		N/C
44 Texas	1965	35	387	1965	35	122	1965	35	170
45 Utah	1980	20	56	1997	3	1	1980	20	40
46 Vermont	Jul-89	11 1/2	8	Jul-89	11 1/2	0	Jul-89	11 1/2	4
47 Virginia	1963	37	106	1963	37	N/A	1963	37	N/A
48 Washington	1978	22	116 b				1983	17	16
49 West Virginia	1967	33	48	NC		NC	NC		NC
50 Wisconsin	1978	22	157	1988	12	5	1978	22	47
51 Wyoming	Apr-79	20 3/4	16	Apr-79	20 3/4	11			
52 Puerto Rico NR									
53 Virgin Islands NR									

a = 1962-65 data reflects partial screening of the birth population; b = includes all clinically significant cases of PKU requiring treatment.

Table 3.12: Total Newborns Screened for Hyperphenylalaninemia

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number of Newborns Confirmed with Classical PKU	Number of Newborns Confirmed w/Clin. Sig. Variant	Number of Newborns Confirmed w/ Variant (Not Clin. Sig.)					
1 Alabama	62,562	7	0	4	N/A	N/A					
2 Alaska	9,866	1	0	0	0	0					
3 Arizona	85,470	44	1	4	2	0					
4 Arkansas	36,840	5	0	0	0	0					
5 California	532,610	188	0	18	21	0					
6 Colorado	65,679	47	0	4	2	DA					
7 Connecticut	43,370	50	0	2	0	6					
8 Delaware	11,639	1	N/A	3	0	1					
9 District of Columbia	15,159	5	2	0	N/A	N/A					
10 Florida	204,305	109	0	15	4	0					
11 Georgia	133,524	0	0	5	2	0					
12 Hawaii	17,638	1	0	0	0	0					
13 Idaho	19,863	10	0	4	0	0					
14 Illinois	181,984	234	1	10	2	7					
15 Indiana	87,891	145	3	10	5	3					
16 Iowa	38,418	16	1	4	0	2					
17 Kansas	39,232	0	0	1	0	0					
18 Kentucky	54,423	9	N/A	4	0	0					
19 Louisiana	68,275	24	0	2	1	0					
20 Maine	13,462	3	0	1	0	0					
21 Maryland	69,574	83	0	2	2	3					
22 Massachusetts	82,673	86	0	1	0	5					
23 Michigan	134,889	197	0	5	4	8					
24 Minnesota	67,546	26	0	3	2	0					
25 Mississippi	42,980	29	0	1	0	0					
26 Missouri	78,302	31	0	7	1	0					
27 Montana	10,927	1	0	0	0	0					
28 Nebraska	24,961	6	1	1	2	2					
29 Nevada	30,387	13	0	2	0	0					
30 New Hampshire	13,987	8	0	0	0	0					
31 New Jersey	112,311	203	0	5	0	5					
32 New Mexico	26,809	36	0	0	N/C	N/C					
33 New York	259,995	205	1	14	11	9					
34 North Carolina	121,347	0	N/D	2	6	ND					
35 North Dakota	8,847	3	0	0	0	0					
36 Ohio	155,943	138	0	9	1	2					
37 Oklahoma	48,650	24	0	2	0	1					
38 Oregon	46,790	20	0	4	1	0					
39 Pennsylvania	146,857	44	0	10	5	3					
40 Rhode Island	13,180	2	0	0	1	0					
41 South Carolina	53,562	26	0	3	0	2					
42 South Dakota	10,589	74	0	0	1	0					
43 Tennessee	84,832	58	0	2	1	N/C					
44 Texas	368,019	0	0	11	4	2					
45 Utah	48,454	46	1	0	0	4					
46 Vermont	6,277	4	0	1	0	1					
47 Virginia	96,755	20	0	1	0	0					
48 Washington	80,453	18	0	6	0	3					
49 West Virginia	21,620	3	0	0	0	0					
50 Wisconsin	68,250	49	0	7	0	2					
51 Wyoming	5,847	1	0	0	0	0					
52 Puerto Rico	59,461	637	101	3	0	0					
53 Virgin Islands	1,851	19	0	0	0	0					
TOTAL	4,125,135	3,009	a	112	a	193	a	81	a	71	a

a = total given does not include all states.

Table 3.13: Summation Results of Testing for Hyperphenylalaninemia

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screening calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary 2nd Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Alabama	62,562	62,173	99.38%			61,927	98.99%		
2 Alaska	9,866	9,821	99.54%			7,759	78.64%	215	2.18%
3 Arizona	85,470	81,956	95.89%			57,352	67.10%		
4 Arkansas	36,840	36,446	98.93%					558	1.51%
5 California	532,610	527,297	99.00%					3,372	0.63%
6 Colorado	65,679	63,219	96.25%	60,530	92.16%				
7 Connecticut	43,370	43,723	* 100.81%			43,942	* 101.32%		
8 Delaware	11,639	11,714	* 100.64%	10,692	91.86%				
9 District of Columbia	15,159	15,125	99.78%					287	1.89%
10 Florida	204,305	204,030	99.87%						
11 Georgia	133,524	189,498	* 141.92%						
12 Hawaii	17,638	17,612	99.85%					187	1.06%
13 Idaho	19,863	20,254	* 101.97%			13,804	69.50%	199	1.00%
14 Illinois	181,984	188,164	* 103.40%						
15 Indiana	87,891	87,639	99.71%					31,782	36.16%
16 Iowa	38,418	38,141	99.28%					1,439	3.75%
17 Kansas	39,232	39,031	99.49%						
18 Kentucky	54,423	54,515	* 100.17%						
19 Louisiana	68,275	67,843	99.37%						
20 Maine	13,462	13,341	99.10%						
21 Maryland	69,574	72,390	* 104.05%	61,152	87.89%				
22 Massachusetts	82,673	82,703	* 100.04%					8,818	10.67%
23 Michigan	134,889	134,022	99.36%						
24 Minnesota	67,546	68,402	* 101.27%					619	0.92%
25 Mississippi	42,980	44,075	* 102.55%						
26 Missouri	78,302	78,640	* 100.43%			13,295	16.98%	5,282	6.75%
27 Montana	10,927	10,825	99.07%			3,550	32.49%		
28 Nebraska	24,961	24,863	99.61%						
29 Nevada	30,387	30,659	* 100.90%	25,668	84.47%				
30 New Hampshire	13,987	13,879	99.23%			815	5.83%		
31 New Jersey	112,311	112,241	99.94%					10,716	9.54%
32 New Mexico	26,809	25,865	96.48%	23,851	88.97%				
33 New York	259,995	258,449	99.41%						
34 North Carolina	121,347	120,750	99.51%						
35 North Dakota	8,847	8,806	99.54%					494	5.58%
36 Ohio	155,943	160,566	* 102.96%					37,021	23.74%
37 Oklahoma	48,650	52,760	* 108.45%						
38 Oregon	46,790	46,879	* 100.19%	42,423	90.67%			861	1.84%
39 Pennsylvania	146,857	148,597	* 101.18%						
40 Rhode Island	13,180	13,150	99.77%					427	3.24%
41 South Carolina	53,562	52,176	97.41%					3,657	6.83%
42 South Dakota	10,589	10,760	101.61%			1,583	14.95%	102	0.96%
43 Tennessee	84,832	79,539	93.76%						
44 Texas	368,019	355,100	96.49%	348,132	94.60%			1,919	0.52%
45 Utah	48,454	47,423	97.87%	46,460	95.88%			387	0.80%
46 Vermont	6,277	6,040	96.22%					326	5.19%
47 Virginia	96,755	99,410	* 102.74%					12,644	13.07%
48 Washington	80,453	76,886	95.57%			69,588	86.50%	1,150	1.43%
49 West Virginia	21,620	21,321	98.62%						
50 Wisconsin	68,250	66,614	97.60%			1,196	1.75%	1,142	1.67%
51 Wyoming	5,847	5,480	93.72%					3,413	58.37%
52 Puerto Rico	59,461	57,155	96.12%					533	0.90%
53 Virgin Islands	1,851	1,851	100.00%			40	2.16%	29	1.57%
TOTAL a	4,125,135	4,159,818	b	618,908	b	274,851	b	127,579	b

* Percentage > 100% denotes inability of program to separate infants screened from specimens received; **a** = totals should be viewed as a rough estimate due to inability of many states, to know actual number of infants screened. Many programs count samples received but cannot eliminate duplication when multiple samples are received; **b** = cannot be calculated due to inaccurate data.

4. Hypothyroidism

4.0 Introduction

Screening for congenital hypothyroidism became a part of many U.S. screening programs in the late 1970's. Initially most U.S. programs utilized screening for thyroxin (T4) as the laboratory screening method of choice. From an initial group of abnormal T4 results, a certain group (usually a group approximating 10% of the total analyzed) was chosen for reanalysis with thyrotropin (TSH). From these two test results newborns were placed in risk categories for follow-up with the most active follow-up being applied to those with low T4 values coupled with high TSH values.

Over the years, as the commercial reagents have become more sensitive and specific, there has been a slow migration of programs towards using a primary TSH testing protocol, but this has been confounded by the tendency among most newborn nurseries to discharge babies earlier, many within the first 12-24 hours. This may result in an increase higher number of false positive TSH reports on newborns screened within the first 12 hours and this must be a consideration of programs considering the primary TSH screening approach. As programs have matured, so, too, have their data and this is reflected in the tables in this section.

4.1 Definitions for Primary Hypothyroidism

Programs were asked to give the definition used by their program to define primary hypothyroidism. There is still not uniform agreement among programs as to the definition to be used for primary hypothyroidism. Most programs use a combination of a low blood spot T4 and elevated TSH, but the definitions of 'high' or 'low' vary. An abnormal screening test is usually followed by a serum thyroid profile interpreted by the physician treating the newborn. It is hoped that eventually programs can come to a consensus as to the best definition to use and the extent to which a pediatric endocrinologist should be involved in the final determination. The results of the survey of definitions used for primary hypothyroidism are given in Table 4.01.

4.2 Definitions for Secondary Hypothyroidism

In addition to detecting primary hypothyroidism, programs using T4 for initial testing have the capability to detect cases of secondary hypothyroidism. Rather than offer a definition, the questionnaire asked programs to give their definition for secondary hypothyroidism for comparison with others. A large number of responders chose to leave this definition to physicians making the diagnosis but a few gave guidance to be considered. Table 4.02 gives the results of this survey.

4.3 Definitions for Transient Hypothyroidism

Programs were asked to give their definition of transient hypothyroidism since this terminology was often used in reports of data submitted for this Report. The results of this request are given in Table 4.03.

4.4 Definitions for 'Other' Types of Hypothyroidism

It was also necessary to request the program's definition for 'other' types of hypothyroidism that might be included. Tertiary hypothyroidism data and other data that might be collected through hypothyroid screening programs. The responses to this question are given in Table 4.04.

4.5 Laboratory Techniques

While most U.S. programs began their hypothyroid screening programs with radioimmunoassay for T4, other techniques are available and have become more popular in some programs, particularly in those using TSH as the primary screening assay. In order to monitor trends among programs in laboratory techniques, programs were asked to report on their laboratory protocols. These data are reported in Table 4.05.

4.6 Initial Screening Results

In order to ascertain the effectiveness of screening for hypothyroidism, programs were asked to report their findings on initial screening. Initial screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of newborns screened and confirmed versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated. These data are reported in Table 4.06.

Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. Programs wishing to explain some of their responses provided information included as footnotes to the table. Due to concerns that some programs may report inaccurate or inappropriate data in some of their responses, no totals were given in some columns of the table. Care should be taken in using these data, since not all programs reported data.

4.7 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are reported in Table 4.07 and are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when a second test is performed without regard for the initial screening result. Programs that requested a second screen because of either a certain result on the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as 'repeat screens' and these are also included.

For each case, the data requested included the total newborns tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. Once again, the reader is cautioned about using the data without regard to notes of explanation.

4.8 Cases of Primary Hypothyroidism Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of primary hypothyroidism, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 4.08. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

4.9 Cases of Secondary Hypothyroidism Divided by Sex and Race/Ethnicity

Programs were also asked to report all diagnosed cases of secondary hypothyroidism and these data are reported in Table 4.09.

4.10 Cases of Transient Hypothyroidism Divided by Sex and Race/Ethnicity

Programs also reported totals of cases of transient hypothyroidism divided by sex and race/ethnicity. These data are reported in Table 4.10.

4.11 Cases of Unclassified Types of Hypothyroidism Divided by Sex and Race/Ethnicity

A limited number of programs reported data on unclassified types of hypothyroidism divided by sex and race/ethnicity. These data are reported in Table 4.11

4.12 Time Until Treatment

In order to look at overall program efficiency, programs were asked to report the number of days from birth until treatment of primary hypothyroidism. The general definition of treatment used in the questionnaire was “initiation of dietary or drug regimen” and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 4.12.

that more meaning can be associated with the data. In particular, the data in this table seem to point out the difficulty of many programs in separating total samples received from total births screened (i.e. many cannot differentiate between first tests received and others that might have been submitted as independent second tests or repeat screens).

4.13 Historic Data

In order to document the value of screening for hypothyroidism, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of various types of cases detected over the years. These data are given in Table 4.13. Since many programs have historically not maintained these data, the reader is cautioned about using them out of context.

4.14 Total Cases Detected

Table 4.14 gives a summation of the data contained in Tables 4.06 and 4.07 along with the number of births in each state or territory as reported in Table 1.01. This table was included so that the reader could view a tabulation of these data without having to refer to several tables.

4.15 Summation of Testing

Table 4.15 is a table summing the significant testing data from this Chapter. It includes the total births from Table 1.01, the total newborns reported as being screened, and the testing data reported in Tables 4.06 and 4.07. Comparative percentages have been provided at the request of a number of readers, but in many cases these data are based on estimates or on data that appear to contain duplicate information thus causing total compliance percentages to exceed 100%. Data responders are requested to review these data and adjust their responses to future questionnaires so

Table 4.01: Definitions for Primary Hypothyroidism

State/Territory	Definition for Primary Hypothyroidism
1 Alabama	Low or Low Normal T4 with TSH elevated ≥ 60 μ IU/mg and confirmed by thyroid profile.
2 Alaska	Low thyroid hormone due to abnormality of the thyroid gland.
3 Arizona	It is the inability of the thyroid to secrete enough T4 with an elevated TSH.
4 Arkansas	Elevated TSH on initial screen, persisting on subsequent testing.
5 California	Elevated TSH, confirmatory testing and designation by MD.
6 Colorado	Diagnosis made by primary care provider and/or pediatric endocrinologist.
7 Connecticut	Classified by treatment center.
8 Delaware	Refer infants with TSH > 50 μ IU/mL; discuss infants T4 < 4 mg/dL and normal TSH. Low BW discuss w/neonatologist.
9 District of Columbia	Elevated TSH.
10 Florida	A permanent pathologic state characterized by abn. low blood levels of thyroid hormones or other as defined by endocrinologists.
11 Georgia	Low or normal T4 with persistent elevation of TSH/confirmed by Endocrinologist with further studies.
12 Hawaii	Low thyroid hormone due to the thyroid gland. Diagnosis made by medical consultant.
13 Idaho	Low thyroid hormone due to an abnormality of the thyroid gland.
14 Illinois	Diagnosis and classification determined by pediatric endocrinologist.
15 Indiana	Low T4, elevated TSH. Diagnosis & classification made by clinical consultant, supported by diagnostic tests.
16 Iowa	Low thyroid hormone levels and high TSH.
17 Kansas	Abnormally low thyroid hormone levels secondary to thyroid dygeneses or defect.
18 Kentucky	Persistent elevations of TSH usually associated with low levels of serum T4.
19 Louisiana	Patient requiring thyroid replacement medication for adequate thyroid functioning.
20 Maine	Persistently elevated TSH & low T4. Confirmed by medical consultant.
21 Maryland	$T4 \leq 6.5$ μ g/dL or 2 SD below mean for run (tray), TSH > 30 μ IU/mL. (Serum total & free are low, TSH elevated)
22 Massachusetts	Classification dependent on endocrinologist's definition.
23 Michigan	TSH elevated ≥ 3 from normal if T4 is normal. TSH elevated ≥ 2 from normal if T4 is low.
24 Minnesota	TSH > 60 μ IU/mL. Final diagnosis made by pediatric endocrinologist.
25 Mississippi	Defined by individual endocrinologist.
26 Missouri	Diagnosis & classification determined by medical consultant.
27 Montana	Elevated TSH (> 40 μ IU/mL) and low or normal T4.
28 Nebraska	Determination made by primary care physician or endocrinologist.
29 Nevada	$T4 > 3$ μ IU/mL regardless of TSH results (FT infants); T4 in lower 3% on 2 specimens.
30 New Hampshire	Classification dependent on endocrinologist's definition.
31 New Jersey	Persistent low T4 levels associated w/highly elevated TSH levels supported by confirmatory serum thyroid profile.
32 New Mexico	Persistently elevated TSH ≥ 40 μ IU/mL; classification of disease may be confirmatory lab and medical consultant.
33 New York	Diagnosis and classification determined by treatment center.
34 North Carolina	Low T4, elevated TSH, serum confirmation, diagnosis and classification made by medical consultant.
35 North Dakota	Persistently elevated TSH & low T4 supported by confirmatory diagnostic laboratory test.
36 Ohio	Diagnosis and classification made by medical consultant.
37 Oklahoma	$T4 < 6$ mg/dL, TSH > 100 μ IU/mL or physician diagnosis.
38 Oregon	Low thyroid hormone due to an abnormality of the thyroid gland.
39 Pennsylvania	Results from an inadequate production of thyroid hormone.
40 Rhode Island	Classification dependent on endocrinologist's definition.
41 South Carolina	Determined by specialist.
42 South Dakota	Diagnosis and classification made by medical consultant.
43 Tennessee	Defined by individual endocrinologist.
44 Texas	Low or normal T4, high TSH; confirmed by serum testing.
45 Utah	Elevated TSH; low T4, T3 RU & FTI.
46 Vermont	$T4 < 5$ μ g/dL; TSH > 100 μ IU/mL; absent/ectopic thyroid gland or insensitivity to TSH.
47 Virginia	Elevated TSH > 60 μ IU/mL with or without low T4 and confirmed by serum thyroid profile.
48 Washington	Persistent elevation of TSH supported by confirmatory diagnostic testing.
49 West Virginia	TSH ≥ 50 mg/dL, confirmed by serum testing and pediatric endocrinologist.
50 Wisconsin	Diagnosis and classification made by treatment center.
51 Wyoming	Determined by primary care provider.
52 Puerto Rico	Infant with T4 < 4.5 μ g/dL & TSH > 10 μ IU/mL on confirmatory testing; infants with serum TSH levels 5-10 μ IU/mL are considered suspect of having primary hypothyroidism.
53 Virgin Islands	$T4 < 7$ μ g/dL, TSH > 40 μ IU/mL.

Table 4.02: Definitions for Secondary Hypothyroidism

State/Territory	Definition for Secondary Hypothyroidism
1 Alabama	Persistent low T4 with normal or slightly elevated TSH and confirmed by thyroid profile.
2 Alaska	It is the inability of the pituitary gland to secrete adequate TSH to stimulate the thyroid (hypothyroid with low TSH).
3 Arizona	It is the inability of the pituitary gland to secrete adequate TSH to stimulate the thyroid (hypothyroid with low TSH).
4 Arkansas	T4, "normal" TSH results not tracked by program.
5 California	Variant hypothyroidism not requiring treatment.
6 Colorado	NC
7 Connecticut	Classified by treatment center.
8 Delaware	Refer infants with TSH > 50 µIU/mL; discuss infants T4 < 4 mg/dL and normal TSH. Low BW discuss w/neonatologist.
9 District of Columbia	Normal TSH, low free T4.
10 Florida	DA
11 Georgia	Low T4 with normal TSH and low T3 uptake/usually confirmed by endocrinologist with further studies.
12 Hawaii	Low thyroid hormone due to an abnormality of the pituitary or hypothalamus. Diagnosis made by medical consultant.
13 Idaho	Low thyroid hormone due to an abnormality of the pituitary or hypothalamus.
14 Illinois	Diagnosis and classification determined by pediatric endocrinologist.
15 Indiana	Diagnosis & classification made by medical consultant, indicated by low T4's.
16 Iowa	Low thyroid hormone level and low TSH.
17 Kansas	Abnormally low thyroid hormone levels secondary to anterior pituitary or hypothalamus dysfunction.
18 Kentucky	Persistence of subnormal levels of free T4, absence of elevations of TSH.
19 Louisiana	DA
20 Maine	Persistent low T4 with normal or slightly elevated TSH. Confirmed by medical consultant.
21 Maryland	Low T4, normal TSH. (Serum total and free are low, serum TSH is low to normal)
22 Massachusetts	Classification dependent on endocrinologist's definition.
23 Michigan	Cause of hypothyroidism is at the level of the pituitary gland. Low T4, normal or low TSH.
24 Minnesota	Classification determined by pediatric endocrinologist.
25 Mississippi	Defined by individual endocrinologist.
26 Missouri	Diagnosis & classification determined by medical consultant.
27 Montana	Elevated TSH (< 40 µIU/mL) and low or normal T4.
28 Nebraska	DA
29 Nevada	T4 in lower 10% and TSH > 100 µIU/mL.
30 New Hampshire	Classification dependent on endocrinologist's definition.
31 New Jersey	Low T4, normal or mildly elevated TSH level, confirmed by thyroid profile and TRH simulation test.
32 New Mexico	Classification determined by consulting endocrinologist.
33 New York	Determination of disease classification made by treatment center.
34 North Carolina	DA
35 North Dakota	Low thyroid hormone level and low TSH.
36 Ohio	Diagnosis & classification made by medical consultant.
37 Oklahoma	NA
38 Oregon	Low thyroid hormone due to an abnormality of the pituitary or hypothalamus.
39 Pennsylvania	DA
40 Rhode Island	Classification dependent on endocrinologist's definition.
41 South Carolina	Determined by specialist.
42 South Dakota	Diagnosis and classification made by medical consultant.
43 Tennessee	Defined by individual endocrinologist.
44 Texas	Low T4, low TSH; confirmed by serum testing.
45 Utah	Low T4, T3 RU, FTI, TSH may be low or normal.
46 Vermont	TBG deficiency (T4 < 5 µg/dL; TSH < 20 µIU/mL; TBG > 1.1); TSH receptor deficit.
47 Virginia	Persistent low T4 with normal or slightly elevated TSH; confirmed by serum thyroid profile.
48 Washington	Classification not made by program.
49 West Virginia	Consistent low T4 (< 4 mg/dL) with normal TSH confirmed by pediatric endocrinologist.
50 Wisconsin	Diagnosis and classification made by treatment center.
51 Wyoming	NC
52 Puerto Rico	Hypothyroidism secondary to pituitary or hypothalamic problem. Diagnosis is made when T4 < 5 with normal TSH and normal TBG.
53 Virgin Islands	Defined by individual endocrinologist.

Table 4.03: Definitions for Transient Hypothyroidism

State/Territory	Definition for Transient Hypothyroidism
1 Alabama	NR
2 Alaska	Low thyroid hormone which spontaneously resolves.
3 Arizona	It is hypothyroidism that ultimately resolves, typically in the first 6 months.
4 Arkansas	Elevated TSH on initial and repeat test, which is subsequently documented to normalize in the absence of treatment.
5 California	Elevated TSH resolves to normal range without treatment.
6 Colorado	NC
7 Connecticut	Classified by treatment center.
8 Delaware	Refer infants with TSH > 50 µIU/mL; discuss infants T4 < 4 mg/dL and normal TSH. Low BW discuss w/neonatologist.
9 District of Columbia	Elevated TSH on initial test, normal TSH & free T4 on subsequent tests when on no thyroxine therapy.
10 Florida	DA
11 Georgia	Initially low or normal T4 with elevated TSH confirmed by serum testing which returns to normal in absence of treatment.
12 Hawaii	Low thyroid hormone which spontaneously resolves. Diagnosis made by medical consultant.
13 Idaho	Low thyroid hormone which spontaneously resolves.
14 Illinois	Diagnosis and classification determined by pediatric endocrinologist.
15 Indiana	Elevated TSH levels that normalize over time with no treatment.
16 Iowa	Low thyroid hormone levels that are corrected by 2-4 weeks of age.
17 Kansas	Temporary decreased thyroid hormone levels and a temporary elevated TSH.
18 Kentucky	DA
19 Louisiana	DA
20 Maine	Low T4 or elevated TSH that returns to normal levels. Evaluation by medical consultant.
21 Maryland	Term babies with a consistently low T4 & mildly elevated TSH for several weeks followed by a return to normal.
22 Massachusetts	Classification dependent on endocrinologist's definition.
23 Michigan	Children treated for congenital hypothyroidism but came off treatment in ≤ 3 years.
24 Minnesota	Classification determined by pediatric endocrinologist.
25 Mississippi	Defined by individual endocrinologist.
26 Missouri	Diagnosis & classification determined by medical consultant.
27 Montana	Elevation possibly due to early collection (<24 hours) of specimen.
28 Nebraska	DA
29 Nevada	NR
30 New Hampshire	Classification dependent on endocrinologist's definition.
31 New Jersey	Persistent low T4 and moderately elevated TSH in which TSH normalizes with time.
32 New Mexico	Elevated TSH correcting to normal by 6-8 weeks. Diagnosis made by consulting endocrinologist.
33 New York	Determination of disease classification made by treatment center.
34 North Carolina	DA
35 North Dakota	Low thyroid hormone levels that are corrected by 2-4 weeks of age.
36 Ohio	Diagnosis & classification made by medical consultant.
37 Oklahoma	Filter paper TSH ≥ 50 µIU/mL; serum TSH < 20 µIU/mL.
38 Oregon	Low thyroid hormone which spontaneously resolves.
39 Pennsylvania	DA
40 Rhode Island	Classification dependent on endocrinologist's definition.
41 South Carolina	Determined by specialist.
42 South Dakota	Diagnosis and classification made by medical consultant.
43 Tennessee	Defined by individual endocrinologist.
44 Texas	Low T4, normal or high TSH on first and/or second screen correcting to normal by age 6 weeks.
45 Utah	NR
46 Vermont	T4 < 5 µg/dL; TSH < 20 µIU/mL for several days/wks; then T4 increases to > 5 µg/dL; often assoc. w/permanency.
47 Virginia	Demonstration of low T4, elevated TSH at time of screening; follow-up testing normal without treatment.
48 Washington	Elevated levels of TSH due to iatrogenic or unknown factors that return to normal & remain euthyroid without treatment intervention.
49 West Virginia	Confirmed by pediatric endocrinologist.
50 Wisconsin	Diagnosis and classification made by treatment center.
51 Wyoming	NC
52 Puerto Rico	Infant with confirmatory T4 < 4.5 µg/dL and TSH > 10 µIU/mL and subsequent testing has normal values.
53 Virgin Islands	Defined by individual endocrinologist.

Table 4.04: Definitions for "Other" Hypothyroidism

State/Territory	Definition for "Other" Hypothyroidism
1 Alabama	NR
2 Alaska	TBG is a biochemical oddity that shows up with a low T4. The TSH is normal.
3 Arizona	TBG is a biochemical oddity that shows up with a low T4 but the person is actually euthyroid. The TSH is normal.
4 Arkansas	DA
5 California	NC
6 Colorado	NC
7 Connecticut	DA
8 Delaware	Refer infants with TSH > 50 µIU/mL; discuss infants T4 < 4 mg/dL and normal TSH. Low BW discuss w/neonatologist.
9 District of Columbia	Elevated TSH and/or low free T4 on initial or subsequent tests.
10 Florida	DA
11 Georgia	ND
12 Hawaii	May also include those who are undetermined in terms of final diagnosis. Compensated hypothyroidism: normal serum T4 or normal free T4 with persistent slightly elevated TSH and a normal thyroid scan. Diagnosis made by medical consultant.
13 Idaho	Low thyroid hormone not due to an abnormality of the thyroid gland or pituitary hypothalamus.
14 Illinois	Diagnosis and classification determined by pediatric endocrinologist.
15 Indiana	Diagnosis & classification by medical consultant, may include TBG deficiency.
16 Iowa	Low thyroid hormone, low TSH, and normal free T4, low TBG.
17 Kansas	DA
18 Kentucky	DA
19 Louisiana	DA
20 Maine	DA
21 Maryland	Low T4 ($\leq 6.5 \mu\text{g/dL}$ or 2 SD below mean for run); TSH $\leq 30 \mu\text{IU/mL}$, usually premature babies.
22 Massachusetts	Classification dependent on endocrinologist's definition.
23 Michigan	Without CH who had normal serum T4 or Free T4 but persistently elevated TSH. Infants with Down Syndrome treated for CH.
24 Minnesota	Classification determined by pediatric endocrinologist.
25 Mississippi	Defined by individual endocrinologist.
26 Missouri	Diagnosis & classification determined by medical consultant.
27 Montana	Low T4 with normal TSH (including TBG, pituitary gland problems and prematurity).
28 Nebraska	DA
29 Nevada	NR
30 New Hampshire	Classification dependent on endocrinologist's definition.
31 New Jersey	Low T4 and normal or borderline abnormal, TSH elevations supported by confirmatory diagnostic test.
32 New Mexico	Diagnosis determined by consulting endocrinologist.
33 New York	Determination of disease classification made by treatment center.
34 North Carolina	DA
35 North Dakota	Low thyroid hormone, low TSH, and normal free T4, low TBG.
36 Ohio	Diagnosis & classification made by medical consultant.
37 Oklahoma	NA
38 Oregon	Low thyroid hormone not due to an abnormality of the thyroid gland or pituitary hypothalamus.
39 Pennsylvania	DA
40 Rhode Island	Classification dependent on endocrinologist's definition.
41 South Carolina	Case records that do not specify type of hypothyroidism.
42 South Dakota	Diagnosis and classification made by medical consultant.
43 Tennessee	Defined by individual endocrinologist.
44 Texas	Low T4 or high TSH; infants who have not yet been diagnosed.
45 Utah	Low TBG, hypothyroxinemia.
46 Vermont	Defects in handling iodine; disorders of hypothalamus/pituitary.
47 Virginia	DA
48 Washington	Classification not made by program.
49 West Virginia	Confirmed by pediatric endocrinologist.
50 Wisconsin	Diagnosis and classification made by treatment center.
51 Wyoming	NC
52 Puerto Rico	Secondary hypothyroidism; infant with T4 level < 4.5 µg/dL, normal TSH, normal TBG, and positive antibodies.
53 Virgin Islands	Defined by individual endocrinologist.

Table 4.05: Hypothyroidism - Laboratory Testing

State/Territory	Primary Screen T4 or TSH	Testing Method for:		Other	NOT NORMAL Definition (T4 = µg/dL; TSH = µIU/mL) <i>Requiring Further Filter Paper Testing</i>	NOT NORMAL Definition (T4 = µg/dL; TSH = µIU/mL) <i>Requiring Immediate Serum Follow-up</i>
		T4	TSH			
1 Alabama	T4	RIA	RIA		T4 < 5.05 µg/dL; TSH >25 µIU/mL	T4 < 5.05 µg/dL; TSH >60 µIU/mL
2 Alaska	T4	RIA	RIA		T4 < 3 µg/dL or TSH > 35 µIU/mL	TSH > 200 µIU/mL
3 Arizona	T4	FIA	FIA		T4 ≤ 6 µg/dL (1st screen) ≤ 5 (2nd screen); TSH ≥ 30 < 60 (1st screen) and ≥ 20 < 60 µIU/mL (2nd screen)	TSH ≥ 60 µIU/mL
4 Arkansas	TSH	FIA	FIA		TSH ≥ 25 - 50 µIU/mL (regardless of T4)	TSH ≥ 50 µIU/mL
5 California	TSH		RIA		TSH 25 - 100	TSH ≥ 100 µIU/mL
6 Colorado	T4	FIA	FIA		T4 < 6 mg/dL and/or TSH ≥ 20 < 40 µIU/mL	T4 any value; TSH ≥ 40 µIU/mL
7 Connecticut	TSH		RIA		TSH ≥ 50 & < 100	TSH ≥ 100
8 Delaware	T4	EIA	EIA		T4 ≤ 4 mg/dL lowest 10%	TSH > 50 µIU/mL
9 District of Columbia	TSH		FIA		TSH ≥ 25 µIU/mL	TSH ≥ 50 µIU/mL
10 Florida	T4	TRIF a	TRIF a		T4 ≤ 6.5 µg/dL; TSH 21-60 µIU/mL	T4 ≤ 6.5 µg/dL; TSH > 60 µIU/mL
11 Georgia	T4	RIA	RIA		T4 ≥ 2500g 5-7.5 <1 wk; 5-5.4 >1 wk; <2500g ≤ 4.5 any age. TSH ≥ 2500g 30-99 <24hrs; 30-74 <48hrs, 25-49 >48; <2500 g <150.	≥ 2500 g = T4 < 5, TSH ≥ 100 @ < 24 hrs, ≥ 75 @ < 48, ≥ 50 > 48. ≤ 2500 g = TSH ≥ 150
12 Hawaii	T4	RIA	RIA		T4 lower 10%; TSH > 35 µIU/mL	T4 lowest 10%; TSH > 100 µIU/mL
13 Idaho	T4	RIA	RIA		T4 < 3 µg/dL or TSH > 25 µIU/mL	T4 < 3 µg/dL; TSH > 60 µIU/mL
14 Illinois	TSH	FIA	FIA		TSH > 54 µIU/mL; T4 5-8 µIU/mL	TSH top 3% but > 54 µIU/mL; T4 < 5 µIU/mL
15 Indiana	T4/TSH	FIA	FIA		T4 < 5.0; TSH 25.5 - 49.9 (>48hrs), 25.5-64.9 (<48hrs)	TSH ≥ 50 (>48hrs) ≥ 65 (<48hrs)
16 Iowa	T4		FIA		TSH ≥ 25 but < 60 µIU/mL	TSH ≥ 60
17 Kansas	T4	RIA	RIA		T4 lowest 10%; TSH ≥ 20 < 60 µIU/mL	T4 lowest 10%; TSH > 60
18 Kentucky	NR	RIA	RIA		T4 < run cutoff, TSH > 20-29; age < 28 days T4 < 6.8 TSH < 20 age ≥ 28 days T4 < 4.0 TSH < 20	T4 < run cutoff; TSH > 30; T4 low 2nd time TSH low > 28 days T4 < 4.0, < 28 days T4 < 6.8
19 Louisiana	T4	EIA	FIA		T4 < 6 µg/dL if ≤ 48 hrs; < 4 µg/dL if > 48 hrs; TSH > 30 µIU/mL	T4 < 6 µg/dL if ≤ 48 hrs; < 4 µg/dL if > 48 hrs; TSH 80 IU/mL
20 Maine	T4	RIA	RIA		T4 < 5 µg/dL; TSH ≥ 20 µIU/mL	
21 Maryland	T4	RIA	EIA		T4 < 6.5 µg/dL or 2 SD below mean for run; TSH > 30 µIU/mL but < 150 µIU/mL if screened < 48 hours	T4 < 6.5 µg/dL or 2SD below mean for run; TSH > 150 µIU/mL if < 48 hrs; > 30 µIU/mL if > 2 days old
22 Massachusetts	T4	FIA	FIA		T4 < 5 µg/dL, TSH ≥ 15 µIU/mL if > 96 hrs. of age TSH ≥ 20 µIU/mL if 24-96 hrs of age TSH ≥ 25 µIU/mL if < 24 hrs of age	TSH - All elevations are reported by phone.
23 Michigan	T4/TSH	FIA	FIA		T4 ≤ 5.0; TSH 23 - 49	TSH ≥ 50
24 Minnesota	TSH		FIA		TSH ≥ 30 µIU/mL but < 60 µIU/mL	TSH > 60 µIU/mL
25 Mississippi	TSH	RIA	FIA		T4 > 4 µg/dL; TSH 20-30 µIU/mL	T4 any value; TSH > 30 µIU/mL blood
26 Missouri	T4/TSH	RIA	RIA		T4 < 6 mg/dL filter paper follow-up: < 24 hr coll. TSH 30-59 > 24 hr. coll. TSH 25-49	TSH ≥ 60 < 24 hr coll.; TSH ≥ 50 > 24 hrs. collections
27 Montana	T4	Fluor.	Fluor.		T4 < 6 µg/dL; TSH ≥ 20 µIU/mL	T4 < 6.0 µg/dL; TSH ≥ 40 µIU/mL
28 Nebraska	T4 / TSH	EIA	EIA			T4 lowest 10%; TSH ≥ 20 µIU/mL
29 Nevada	T4	RIA	RIA		T4 < 3 µg/dL	T4 low 10% ; TSH > 100 µIU/mL
30 New Hampshire	T4	RIA	RIA		T4 < 5.1 µg/dL, TSH ≥ 15 µIU/mL if > 96 hrs. of age TSH ≥ 20 µIU/mL if 24-96 hrs of age TSH ≥ 25 µIU/mL if < 24 hrs of age	All elevations reported by phone.
31 New Jersey	T4	FIA	FIA		T4 lowest 10%; TSH ≥ 20-40 µIU/mL	T4 lowest 10%; TSH ≥ 40 µIU/mL
32 New Mexico	TSH	FIA			TSH ≥ 25 IU/mL	TSH ≥ 40 µIU/mL
33 New York	T4	RIA	RIA		T4 lowest 10%; TSH > 20-30 µIU/mL	T4 < 2SD below mean; TSH ≥ 20 µIU/mL
34 North Carolina	T4	RIA	IRMA		T4 < 5 µg/dL; TSH ≤ 20 µIU/mL and < 40 µIU/mL	TSH ≥ 40 µIU/mL
35 North Dakota	TSH		FIA		TSH ≥ 25 but < 60 µIU/mL	TSH ≥ 60 µIU/mL
36 Ohio	T4	FIA	FIA			T4 < 8 µg/dL; TSH > 25 µIU/mL
37 Oklahoma	T4	RIA/EIA	RIA/EIA		T4 lower 10% of sample and TSH ≥ 25-50 µIU/mL	T4 lower 10% of sample and TSH ≥ 50 µIU/mL
38 Oregon	T4	RIA	RIA		T4 < 3 µg/dL or TSH > 35 µIU/mL	TSH > 200 µIU/mL
39 Pennsylvania	T4	RIA	RIA IRMA		T4 ≥ 6 µg/dL and TSH ≥ 25 µIU/mL < 35 µIU/mL	T4 ≤ 6.0 µg/dL and TSH ≥ 25 µIU/mL or T4 > 6 & TSH ≥ 35
40 Rhode Island	T4/TSH	FIA	FIA		T4 ≤ 5; TSH < 15 if > 96 hrs, < 20 if 24-96 hrs, < 25 if < 24 hrs	T4 ≤ 3 µg/dL; TSH > 20
41 South Carolina	T4	EIA	EIA		T4 ≥ 2 SD below mean or ≤ 9 µg/dL; TSH ≥ 20 µIU/mL T4 ≤ 9.0 µg/dL ≤ 7 days old; T4 ≤ 4 µg/dL ≥ 8 days old	TSH > 20 µIU/mL; TSH > 30 referred to MD of record.
42 South Dakota	T4	FIA	FIA		T4 < 6 µg/dL; TSH 0-30 µIU/mL	T4 < 6 µg/dL; TSH > 30 µIU/mL
43 Tennessee	TSH		FIA		TSH 33-55 µIU/mL	TSH ≥ 55 µIU/mL
44 Texas		RIA	RIA		T4 lowest 0.5%; TSH elevations up to 99 µIU/mL	T4 low/normal; TSH ≥ 100 µIU/mL
45 Utah	T4	FIA	FIA		T4 ≤ 4.0 µg/dL; TSH ≥ 25 µIU/mL	T4 - 2 abn. screens; TSH > 200 µIU/mL & 2 abn. Screens
46 Vermont	T4	RIA	RIA		T4 ≤ 5 µg/dL and/or TSH > 15 µIU/ml if > 96 hrs. repeat FP: if TSH > 20 µIU/ml if 24-96 hrs. and > 25 µIU/ml if < 24 hrs.	T4 < 5 µg/dL; TSH > 50 µIU/mL
47 Virginia	T4	FIA	FIA		T4 > 5.5 SD below the assay; TSH ≥ 25 µIU/mL	T4=2 consecutive samples w/low T4; TSH ≥ 60 µIU/mL
48 Washington	T4	RIA	FIA		T4 ≤ 10 µg/dL & TSH ≥ 20 µIU/mL or T4 ≥ 10 µg/dL & TSH ≥ 30 µIU/mL	TSH ≥ 100 µIU/mL
49 West Virginia	T4	RIA	RIA		T4 ≤ 6.8 µg/dL	T4 < 4 µg/dL; TSH > 50 µIU/mL
50 Wisconsin	TSH		FIA		TSH 0-25 hrs age ≥ 37 but < 50 µIU/mL 26-96 hrs age ≥ 30 but < 50 µIU/mL	TSH ≥ 50 µIU/mL; 96+ hrs age TSH ≥ 20 µIU/mL
51 Wyoming	T4	FIA	FIA		T4 < 6 mg/dL and/or TSH ≥ 20 < 40 µIU/mL	T4 any value; TSH ≥ 40 µIU/mL
52 Puerto Rico	T4/TSH	RIA	IRMA		T4 6-9 µg/dL; TSH > 20 µIU/mL	T4 < 6 µg/dL; TSH > 20 µIU/mL
53 Virgin Islands		RIA	RIA		T4 > 7 µg/dL; TSH < 25 µIU/mL	T4 > 7 µg/dL; TSH > 40 µIU/mL

a = Time Resolved Immuno Fluorescence; b = If T4 results are not normal (<9.0 µg/dL), serum T4 or free T4 and serum TSH are done. Further filter paper testing on an abnormal screening result is not done.

Table 4.06: Initial Screening Results - Hypothyroidism

State/Territory	Number of Newborns Screened	Number of Newborns with NOT NORMAL Test Results	Number of Newborns with NOT NORMAL Test Results Lost to Follow-up	Number of Newborns Confirmed with Primary Hypothyroidism	Number of Newborns Confirmed with Secondary Hypothyroidism	Number of Newborns Confirmed with Transient Hypothyroidism	Number of Newborns Confirmed w/"Other" Hypothyroidism
1 Alabama	62,173	0	0	10	0	0	0
2 Alaska	9,821	103	0	1	0	0	0
3 Arizona	81,956	735	1	37	1	27	8 k
4 Arkansas	36,456	566	6	13	0	0	0
5 California	527,297	1,046	4	254	10	3	0
6 Colorado	63,219	549 a	6 b	19	NC	NC	NC
7 Connecticut	43,728	37	0	12	0	0	0
8 Delaware	11,714	13	NA	7	1	0	5
9 District of Columbia	15,125	84	69 j	1	NA	0	NA
10 Florida	204,030	780	0	65	0	2	0
11 Georgia	189,498	0	0	50	6	1	0
12 Hawaii	17,612	83	22 i	11	0	3	0
13 Idaho	20,254	98	0	7	1	0	0
14 Illinois	188,164 d	1,388	0	60	NA	15	2
15 Indiana	87,639	2,585	231	26	0	3	0
16 Iowa	38,141	282	10	25	NA	NA	NA
17 Kansas	39,031	1,537	183	33	ND	1	1
18 Kentucky	54,515	335	0	20			
19 Louisiana	67,843	332	4	10	DA	DA	DA
20 Maine	13,341	117	0	9			
21 Maryland	72,390	498	45	9	1	0	5
22 Massachusetts	82,703	1,046	N/A	34	0	1	
23 Michigan	134,022	6,401	106	34	2	6	63
24 Minnesota	68,402	50	0	22	4	5	
25 Mississippi	44,075	30	0	2	0	0	0
26 Missouri	78,640	351	0	34	1	3	0
27 Montana	10,825	88	33	2	0	0	1 c
28 Nebraska	24,863	114	2 i	8	DA	DA	DA
29 Nevada	30,659	570	0	8	0	0	0
30 New Hampshire	13,879	75	0	16	0	0	0
31 New Jersey	112,241	1,745	0	60	7	54	23
32 New Mexico	25,745	656	0	16	NC	NC	NC
33 New York	258,449	4,569	NC	151	4	NA	22
34 North Carolina	120,750	2,877	ND	29	DA	DA	DA
35 North Dakota	8,806	115	3 e	3	0	3	0
36 Ohio	160,566	2,492	0	75	1	3	1
37 Oklahoma	52,760	321	14	24	0	8	0
38 Oregon	46,879	641	0	26	0	0	0
39 Pennsylvania	145,874	0	0	46	0	0	0
40 Rhode Island	13,150	0	0	4	0	2	0
41 South Carolina	52,176	3,016	284	23	0	11	13
42 South Dakota	10,582	67	0	0	0	0	0
43 Tennessee	79,539	448	3	59	NC	NC	NC
44 Texas	355,100	5,022	142	157	0	27	4
45 Utah	47,423	220	6	15	3	5	10
46 Vermont	6,040	67	0	3	0	64	0
47 Virginia	99,410	1,216	0	18	0	9	1 f
48 Washington	76,886	401	3	18	NA	2	NA
49 West Virginia	21,321	216	ND	6	ND	ND	ND
50 Wisconsin	66,614	238	6	21	DA	3	DA
51 Wyoming	5,480	46	2	2	0	0	0
52 Puerto Rico	57,155	1,096	253	6	0	75	0
53 Virgin Islands	1,851	13	0	0	0	0	0
TOTAL	g	g	g	1,601 h	42 h	336 h	159 h

a = 474 borderline, 75 presumptive positive; **b** = certified letters to parents, no further information; **c** = TBG; **d** = includes repeat specimens; **e** = 1 clinic not responding to follow-up letters, 1 moved out of state, 1 parent refused retest; **f** = hypopituitarism; **g** = totals not given - too many programs reported inaccurate or inappropriate data; **h** = total given does not include all states; **i** = expired; **j** = lab lost to followup-DC MCH has more info; **k** = 7 TBG def, 1 hyperthyroidism.

Table 4.07: Second Screens for Hypothyroidism

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen R	Newborns Receiving Discretionary Second Screens D	Newborns Receiving Repeat Second Screens P	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed w/primary Hypo- thyroidism				Number of Newborns Confirmed w/secondary Hypo- thyroidism				Number of Newborns Confirmed w/transient Hypo- thyroidism				Number of Newborns Confirmed w/other Hypo- thyroidism					
					R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total		
1 Alabama		62,731	420	63,151																										
2 Alaska		7,759	313	8,072		48	10																3	b	3					
3 Arizona		57,352		57,352		106		106				0		3		3					14		14			10		10	f	
4 Arkansas			DA	567	567		DA	17	17		DA		0		DA		0		DA	0	DA		DA	0	0		DA	0	0	
5 California			3,392	3,392				0				0	0				0					0	0							
6 Colorado	60,530		N/C		4			N/C	4				N/C	N/C	1				N/C	1	N/C	N/C		N/C	N/C		N/C	N/C		
7 Connecticut			290	290		0	11	11		0		0		0		0		0		0	0	0		0	0		0	0		
8 Delaware	10,692			10,692				0	N/A		N/A	N/A	0		0		0		0		0	0	0		0	0		0	0	
9 District of Columbia			287	287				0				0				0					0	0						0		
10 Florida				0																										
11 Georgia			ND	ND																										
12 Hawaii			DA	247	247		DA	58	58				0	0				0	0					0	0			0	0	
13 Idaho		13,804	202	14,006		13	6	19						1		1														
14 Illinois			N/C	0																										
15 Indiana			DA	31,782	31,782			472	472				38	38				0					0					0		
16 Iowa			1,439	1,439				0		DA	0	0		DA	0	0		DA	0	0		DA	0	0		DA	0	0		0
17 Kansas				0				0				0				0					0						0			
18 Kentucky	NR																													
19 Louisiana				0																										
20 Maine																														
21 Maryland	61,150			61,150	313			313	56			56	6			6										44		44		
22 Massachusetts			8,818	8,818				228	228				0			4	4		1	1					0			0		
23 Michigan				0				0	0																					
24 Minnesota			887	887				342	342																					
25 Mississippi		0		0		0		0		0	0	0		0	0	0		0	0	0		0	0	0		0	0	0		0
26 Missouri		13,295	5,282	18,577		120	51	171								0														

continued

Table 4.07: Second Screens for Hypothyroidism (continued)

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed w/primary Hypo- thyroidism				Number of Newborns Confirmed w/secondary Hypo- thyroidism				Number of Newborns Confirmed w/transient Hypo- thyroidism				Number of Newborns Confirmed w/other Hypo- thyroidism			
	R	D	P		R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total
27 Montana		3,543		3,543																								
28 Nebraska		DA	196	196		DA	DA																					
29 Nevada	25,668			25,668	142		0	142																				
30 New Hampshire		815		815																								
31 New Jersey		DA	10,716	10,716		DA	773	773		DA		0		DA		0		DA		0		DA		0		DA		0
32 New Mexico	23,581		168	23,749				0																				
33 New York		DA		0		DA	N/C			DA	N/C			DA	N/C			DA	N/C			DA	N/C			DA	N/C	
34 North Carolina		N/C	a			N/C	N/C			N/D	N/D		1	0	1		DA	DA	DA		DA	DA	DA		DA	DA	DA	
35 North Dakota		DA	494	494		DA	1	1		DA	0	0		DA	0	0		DA	0	0		DA	0	0		DA	0	0
36 Ohio		N/C	37,021	37,021																								
37 Oklahoma		N/C	N/C			N/C	N/C			N/C	N/C			N/C	N/C			N/C	N/C			N/C	N/C			N/C	N/C	
38 Oregon	43,423		874	44,297	185		44	229																				
39 Pennsylvania																												
40 Rhode Island			427	427																								
41 South Carolina		N/A	3,657	3,657		N/A	N/A	N/A		N/A	N/A	N/A		N/A	N/A	N/A		N/A	N/A	N/A		N/A	N/A	N/A		N/A	N/A	N/A
42 South Dakota		180	51	231		4	4	8					5	5														
43 Tennessee				0																								
44 Texas	348,132		1,919	350,051				0	0			0		0	0	0		0	0	0		0	0	0		0	0	0
45 Utah	46,460		571	47,031	72		72	3			3	8		8	0		0	0	12			12	13				13	
46 Vermont			326	326			3	3																				
47 Virginia			12,708	12,708			403	403		0	0	0		0			0	0			0	0	0		0	0	0	
48 Washington		69,187	1,551	70,738		12	NR	12		0	NR	0		2	NR	2		N/A	NR		0	NR	0		N/A	NR		
49 West Virginia			N/A	0		30	84	114		0		0				0												
50 Wisconsin		1,196	1,284	2,480		6	9	15		0	0	0		3	0	3		DA	DA		0	0	0		DA	DA		
51 Wyoming	DA	DA	N/C			DA	DA	N/C		DA	DA	N/C		DA	DA	N/C		DA	DA	N/C		DA	DA	N/C		DA	DA	N/C
52 Puerto Rico			843	843			6	6																				
53 Virgin Islands		10	20	30																								
Total	c	619,636	229,872	126,752	d			d				d	15	10	9	34 e	0	0	1	1 e	12	14	0	26 e	57	10	3	70 e

a = data requested were not collected by the program therefore number of specimens rather than number of infants was counted; b = TBG; c = totals given reflects program responses, should be viewed as estimates only, given the number not reporting & the number of caveats listed; d = totals not given - too many programs reported inaccurate or inappropriate data; e = total given does not include all states; f = 8 TBG def, 1 TBG excess, 1 other (confirmed CAH).

Table 4.08: Cases of Primary Hypothyroidism

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Total	Hispanic		Total	
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female		Male	Female		
1 Alabama															10 ¹				
2 Alaska		1												0	1	1			
3 Arizona	9	17					1		5	7		1	15	25	40	7	12	19	
4 Arkansas	4	5	1	1							1			5	7	13 ³	1	1	
5 California	20	41	2	8	9	11		1	2	3	3	1	36	65	254 ³	44	109	153	
6 Colorado a												6	14	6	14	20			
7 Connecticut	1	4							2			2	3	5	7	12	1	1	2
8 Delaware	3	4												3	4	7			
9 District of Columbia												1	0	1	1				
10 Florida	14	33	1	4		1					2	11	17	49	65 ²				
11 Georgia	12	20	9	3	1								22	23	50 ³	1	4	5	
12 Hawaii		1		1	6	2			1				7	4	11				
13 Idaho	1	7											1	7	8				
14 Illinois	11	20		2	3	5					1	3	15	30	60 ³	3	12	15	
15 Indiana	12	13			1								13	13	26				
16 Iowa	12	12		1									12	13	25		2	2	
17 Kansas											17	16	17	16	33				
18 Kentucky											6	14	6	14	20				
19 Louisiana	2	4		1					2				1	4	6	10			
20 Maine	6	3											6	3	9				
21 Maryland	3	5	2		1				1			1	8	5	15 ³		2	2	
22 Massachusetts											21	17	21	17	38				
23 Michigan	9	13	2	2		1			1	3		1	12	20	34 ³		2	2	
24 Minnesota	13	6									2	1	15	7	22				
25 Mississippi		1	1										1	1	2				
26 Missouri	15	12	1	2						2	1	1	17	17	34				
27 Montana	1					1							1	1	2				
28 Nebraska	1	4		1				1					1	5	8 ³	2		2	
29 Nevada	1	3											1	3	8 ³	1	3	4	
30 New Hampshire											7	9	7	9	16				
31 New Jersey	10	18	2	9		1			1	1	10	8	23	37	60				
32 New Mexico	1	6					3	3		1	1	1	5	11	16	2	5	7	
33 New York	33	46	16	4	8	2	1	1	3	7			61	60	151 ³	11	19	30	
34 North Carolina	8	13		2	1		1		3	1		1	13	17	30				
35 North Dakota	1	2											1	2	3				
36 Ohio											34	41	34	41	75				
37 Oklahoma	7	9	2	1			1				1	1	11	11	24 ³	1	1	2	
38 Oregon	11	15											11	15	26	2	2	4	
39 Pennsylvania															46 ¹				
40 Rhode Island	1	3											1	3	4				
41 South Carolina	6	11	4	1					1	b			11	12	23	1	0	1	
42 South Dakota															5 ¹				
43 Tennessee	18	25	1	8					2	5			21	38	59				
44 Texas	19	41	3	9					5	6	2		29	56	157 ³	25	47	72	
45 Utah											11	12	11	12	23				
46 Vermont		3											0	3	3				
47 Virginia	3	8	1	3							1	2	5	13	18	1	2	3	
48 Washington	2	8	1	1	3	1		1		2		1	6	14	20		5	5	
49 West Virginia	4	2											4	2	6				
50 Wisconsin	5	16		1	2								7	17	24				
51 Wyoming	1	1											1	1	2				
52 Puerto Rico									3	3			3	3	6	3	3	6	
53 Virgin Islands																			
TOTAL	280	456	49	65	35	25	7	6	32	42	129	161	532	755	1,635	105	232	337	

¹did not report a breakdown of Race/Ethnicity.

²total reported is from Table 4.06 and Table 4.07; therefore total does not equal or may differ from data given in Table 4.08 and denotes a problem in reliability.

³Ethnicity counted as a Race and is included in total.

a = race/ethnicity data not collected; **b** = no race given, only marked Hispanic.

Table 4.09: Cases of Secondary Hypothyroidism

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
1 Alabama															NA			
2 Alaska													0	0	0			
3 Arizona									1				1	0	1	1		1
4 Arkansas																		
5 California		2	1			4						1	6	10 ³	1	2	3	
6 Colorado															NC			
7 Connecticut																		
8 Delaware		1											0	1	1			
9 District of Columbia																		
10 Florida																		
11 Georgia	1	2	1	2									2	4	6			
12 Hawaii																		
13 Idaho	1												1	0	1			
14 Illinois															NA			
15 Indiana															DA			
16 Iowa																		
17 Kansas																		
18 Kentucky																		
19 Louisiana															DA			
20 Maine																		
21 Maryland	1												1	0	1			
22 Massachusetts												1	0	1	1			
23 Michigan		1	1										1	1	2			
24 Minnesota	1	2				1							1	3	4			
25 Mississippi																		
26 Missouri	1												1	0	1			
27 Montana																		
28 Nebraska															DA			
29 Nevada																		
30 New Hampshire																		
31 New Jersey	2		1	2									2	3	4	7		
32 New Mexico															NC			
33 New York		2	1						1				2	2	4			
34 North Carolina															DA			
35 North Dakota															0			
36 Ohio													1	0	1			
37 Oklahoma																		
38 Oregon																		
39 Pennsylvania																		
40 Rhode Island																		
41 South Carolina																		
42 South Dakota															NA			
43 Tennessee															NC			
44 Texas																		
45 Utah												3		3	0	3		
46 Vermont																		
47 Virginia																		
48 Washington															NA			
49 West Virginia															ND			
50 Wisconsin															DA			
51 Wyoming																		
52 Puerto Rico																		
53 Virgin Islands																		
TOTAL	7	10	5	4	0	5	0	0	2	0	3	4	17	23	43	2	2	4

¹ did not report a breakdown of Race/Ethnicity.

² total reported is from Table 4.06 and Table 4.07; therefore total does not equal or may differ from data given in Table 4.09 and denotes a problem in reliability.

³ Ethnicity counted as a Race and is included in total

Table 4.10: Cases of Transient Hypothyroidism

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic					
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total		
1 Alabama																			N/A	
2 Alaska															0					
3 Arizona	18	5	2	2	1		1	1	3	8			25	16	41	9	8	17		
4 Arkansas																				
5 California		1			1								1	1	3	3	0	1	1	
6 Colorado																			N/C	
7 Connecticut																				
8 Delaware																				
9 District of Columbia																				
10 Florida															2				a	
11 Georgia			1										1	0	1					
12 Hawaii					1	1			1				2	1	3					
13 Idaho															0					
14 Illinois	4	1		1	1							2	1	7	3	15	3	2	3	5
15 Indiana	1	1		1										1	2	3				
16 Iowa																				
17 Kansas												1		1	0	1				
18 Kentucky																				
19 Louisiana																				DA
20 Maine																				
21 Maryland																				
22 Massachusetts											1		1	0	1					
23 Michigan	4				1									5	0	6	3	1	1	
24 Minnesota	2	2										1		3	2	5				
25 Mississippi															0					
26 Missouri	2		1											3	0	3				
27 Montana																				
28 Nebraska																				DA
29 Nevada																				
30 New Hampshire																				
31 New Jersey	7	1	4	4	1	1			1			21	14	34	20	54				
32 New Mexico																				N/C
33 New York																				N/A
34 North Carolina																				DA
35 North Dakota	1	2												1	2	3				
36 Ohio												2	1	2	1	3				
37 Oklahoma	2	1		2			1	1						3	4	8	3	1	0	1
38 Oregon																				
39 Pennsylvania																				
40 Rhode Island	1	1												1	1	2				
41 South Carolina			8	3										8	3	11				
42 South Dakota																				N/A
43 Tennessee																				N/C
44 Texas																				27 ¹
45 Utah											8	9	8	9	17					
46 Vermont	48	15												48	15	64	2			
47 Virginia	5					1			1	1			1	6	3	9	0	1	1	
48 Washington						2								0	2	2				
49 West Virginia																				N/D
50 Wisconsin	1	1										1		2	1	3				
51 Wyoming																				
52 Puerto Rico																				75 ¹
53 Virgin Islands																				
TOTAL	96	31	16	13	6	5	2	2	6	9	37	26	163	86	362	12	14	26		

¹did not report a breakdown of Race/Ethnicity.

²total reported is from Table 4.06 and Table 4.07; therefore total does not equal or may differ from data given in Table 4.09 and denotes a problem in reliability.

³Ethnicity counted as a Race and is included in total

a = not officially confirmed or treated.

Table 4.11: Cases of Other or Unclassified Hypothyroidism

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Alabama																	
2 Alaska															3 ¹			
3 Arizona	2												2	0	18			
4 Arkansas																		
5 California																		NC
6 Colorado																		NC
7 Connecticut																		
8 Delaware	4		1										5	0	5			
9 District of Columbia																		
10 Florida																		NA
11 Georgia																		
12 Hawaii																		
13 Idaho																		
14 Illinois	1	1											1	1	2			
15 Indiana																		DA
16 Iowa																		
17 Kansas	1												1	0	1			
18 Kentucky																		
19 Louisiana																		DA
20 Maine																		
21 Maryland	2	5	9	8					1	1	12	11	24	25	49			
22 Massachusetts																		
23 Michigan	27	16	3	4		1			3	7	1	40	22	63 ³		1	1	
24 Minnesota																		
25 Mississippi																		
26 Missouri																		
27 Montana	1												1	0	1			
28 Nebraska																		DA
29 Nevada																		
30 New Hampshire																		
31 New Jersey	6	5	4	1	1				2	4		17	6	23				
32 New Mexico																		NC
33 New York															22 ¹			
34 North Carolina																		DA
35 North Dakota																		
36 Ohio										1		1	0	1				
37 Oklahoma																		
38 Oregon																		
39 Pennsylvania																		
40 Rhode Island																		
41 South Carolina	3	2	4	4									7	6	13			
42 South Dakota																		NA
43 Tennessee																		NC
44 Texas						1							0	1	4 ³	2	1	3
45 Utah											19	4	19	4	23			
46 Vermont																		
47 Virginia						1							0	1	1			
48 Washington																		NA
49 West Virginia																		ND
50 Wisconsin																		DA
51 Wyoming																		
52 Puerto Rico																		
53 Virgin Islands																		
TOTAL	47	29	21	17	1	3	0	0	6	1	43	16	118	66	229	2	2	4

¹ did not report a breakdown of Race/Ethnicity.

² total reported is from Table 4.06 and Table 4.07; therefore total does not equal or may differ from data given in Table 4.11 and denotes a problem in reliability.

³ Ethnicity counted as a Race and is included in total.

Table 4.12: Days from Birth Until Treatment Initiated for Primary Hypothyroidism
Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	TOTAL
1 Alabama															10	10
2 Alaska													1			1
3 Arizona	1		3	2	3	1	4	5	1		2		5	13		40
4 Arkansas							1	1	1				4	6		13
5 California	9	21	29	30	22	25	20	16	16	9	6	9	20	18	4	254
6 Colorado				1	1	2		4		1			6	4	1	20
7 Connecticut						2	1	3		3	1		1	1		12
8 Delaware				1	1		1	1	1	1				1		7
9 District of Columbia																1 ¹
10 Florida			1		2	1	1	10	11	4	6	9	10	9	1	65
11 Georgia													13		42	50 ²
12 Hawaii								1	1	1	1		4	2	1 ^a	11
13 Idaho									1			2		5		8
14 Illinois			1		7	1	4	1	5	5	7	3	4	21	1	60
15 Indiana		2			4	5	1	4			1		2	4	3	26
16 Iowa		1		2		3		1			1		1	9	7	25
17 Kansas		1		1		1	1	1	1			2	6	19		33
18 Kentucky															20	20
19 Louisiana	1								1	1			5	2		10
20 Maine					1		1		1		1		5			9
21 Maryland						2			1		1		2	7	2	15
22 Massachusetts															38	38
23 Michigan					1	5	3	4	2	4	2	1	4	8		34
24 Minnesota				3	3	1	5	3		1			5	1		22
25 Mississippi								1						1		2
26 Missouri								4	4	3	2	2	3	15	1	34
27 Montana							1							1		2
28 Nebraska				3	1			3					1			8
29 Nevada	1		1					1				1	3	1		8
30 New Hampshire															16	16
31 New Jersey				1	4	2	10	2	1	3	1	2	5	29		60
32 New Mexico				1	2	1	2	2		1	1	4	1		1	16
33 New York			1	4	9	13	14	7	13	6	5	12	22	45		151
34 North Carolina															30	30
35 North Dakota													2	1		3
36 Ohio															75	75
37 Oklahoma	1				2		1	2		1		1	8	8		24
38 Oregon	1					3	3	3		1	1	1	5	7	1	26
39 Pennsylvania															46	46
40 Rhode Island				1		1								1	1	4
41 South Carolina			1		1	1		1		1		1	3	7	7	23
42 South Dakota															5	5
43 Tennessee															59	59
44 Texas															157	157
45 Utah			1		2	1	2			1		1	5	8	2	23
46 Vermont		1	1									1				3
47 Virginia	1				3	4	1		1	2	1		2	3		18
48 Washington					2	2	2	1	2		1	1	4	5		20
49 West Virginia															6	6
50 Wisconsin		1	1	1	2	3	2		4			1	1	8		24
51 Wyoming															2	2
52 Puerto Rico													6			6
53 Virgin Islands																0

¹did not report a breakdown of Days from Birth.

²total reported is from Table 4.06 and Table 4.07; therefore does not equal or may differ from data given in Table 4.12 and denotes a problem in reliability.

a = infant expired before treatment could be initiated.

Table 4.13: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Primary Hypothyroidism			Secondary Hypothyroidism			Transient Hypothyroidism			Other or Unclassified Hypothyroidism		
	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases
1 Alabama	NR											
2 Alaska	1987	13	47	1987	13	8	N/C		N/C	1987	13	10
3 Arizona	1994	6	211	1994	6	4	1996	4	66	1996	4	81
4 Arkansas	1984	16	119									
5 California	1980	20	3,467	1980	20	97	1980	20	68	DA		DA
6 Colorado	Mar-79	20 3/4	298	DA		DA	DA		DA	DA		DA
7 Connecticut	1976	24	267	N/D		N/D	N/D		N/D	N/D		N/D
8 Delaware	1993	7	21	1993	7	1	1993	7	0	1993	7	29
9 District of Columbia	1996	4	12	1996	4	0	1996	4	1	1996	4	0
10 Florida	1978	22	831	DA		DA	DA		DA	DA		DA
11 Georgia	Sep-78	21 1/4	558	Sep-78	21 1/4	46	Sep-78	21 1/4	30	Sep-78	21 1/4	0
12 Hawaii	1986	14	66	1986	14	7	1986	14	70	1986	14	27
13 Idaho	1976	24	106	1976	24	8						
14 Illinois	1980	20	1,014	N/A		N/A	1992	8	62	1995	5	20
15 Indiana	Jul-85	15 1/2	309				2000	0	3			
16 Iowa	N/C		N/C	N/C		N/C	N/C		N/C	N/C		N/C
17 Kansas	1977	23	187				1999	1	1	1977	23	1
18 Kentucky	NR											
19 Louisiana	1980	20	229	N/C		N/C	DA		DA	DA		DA
20 Maine	1976	24	99	N/C		N/C	N/D		N/D	N/D		N/D
21 Maryland	1979	21	330	1979	21	5	1993	7	3	1993	7	290
22 Massachusetts	1976	24	650 c	N/A		N/A	N/A		N/A	N/A		N/A
23 Michigan				N/A		N/A	N/A		N/A	N/A		N/A
24 Minnesota	NR											
25 Mississippi	NR											
26 Missouri	NR											
27 Montana	N/C		N/C	N/C		N/C	N/C		N/C	N/C		N/C
28 Nebraska	1989	11	86 a	N/C		N/C	N/C		N/C	N/C		N/C
29 Nevada	NR											
30 New Hampshire	1976	24	104	N/C		N/C	N/C		N/C	N/C		N/C
31 New Jersey	NR											
32 New Mexico	1981	19	205	N/C		N/C	N/C		N/C	N/C		N/C
33 New York	1979	21	2,591	1979	21	131				1979	21	469
34 North Carolina	NC		N/C	N/C		N/C	N/C		N/C	N/C		N/C
35 North Dakota	1979	21	51				1999	1	5			
36 Ohio	NR											
37 Oklahoma	1991	9	163				N/C		N/C	N/C		N/C
38 Oregon	NR											
39 Pennsylvania	NR											
40 Rhode Island				N/A		N/A						
41 South Carolina	1996	4	71	1996	4	1	1996	4	96	1996	4	23 b
42 South Dakota	NR											
43 Tennessee	NR											
44 Texas	1980	20	2,381	N/C		N/C	N/C		N/C	N/C		N/C
45 Utah	1980	20	281	1980	20	8	1980	20	177	1980	20	173
46 Vermont	Jul-89	11 1/2	21	Jul-89	11 1/2	2				Jul-89	11 1/2	0
47 Virginia	1978	22	385	1978	22	N/A	1978	22	N/A	1978	22	2
48 Washington	1978	22	409	N/A		N/A	N/C		N/C	N/A		N/A
49 West Virginia	N/D		N/D	N/D		N/D	N/D		N/D	N/D		N/D
50 Wisconsin	1978	22	438	DA		DA	1991	9	45	DA		DA
51 Wyoming	Apr-79	20 3/4	35	Apr-79	20 3/4	2	Apr-79	20 3/4	1	Apr-79	20 3/4	0
52 Puerto Rico	1981	19	NC	1981	19	NC	1981	19	NC	1981	19	NC
53 Virgin Islands	NR											

a = data not collected prior to 1989; b = prior to 1996 all were unclassified, there were 111 from 7/79 through 12/95; c = includes secondary, transient and other hypothyroidism cases.

Table 4.14: Total Newborns Screened for Hypothyroidism

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up	Number of Newborns Confirmed w/Primary Hypo-thyroidism	Number of Newborns Confirmed w/Secondary Hypo-thyroidism	Number of Newborns Confirmed w/Transient Hypo-thyroidism	Number of Newborns Confirmed with Other Hypo-thyroidism
1 Alabama	62,562	N/A	N/A	10	0	0	0
2 Alaska	9,866	161	0	1	0	0	3 a
3 Arizona	85,470	841	1	40	1	41	18
4 Arkansas	36,840	583	6	13	DA	0	0
5 California	532,610	1,046	4	254	10	3	NC
6 Colorado	65,679	553	6	20	N/C	N/C	N/C
7 Connecticut	43,370	48	0	12	0	0	0
8 Delaware	11,639	13	N/A	7	1	0	5
9 District of Columbia	15,159	84	69	1	0 N/A	0	0 N/A
10 Florida	204,305	780	0	65	DA	2	DA
11 Georgia	133,524	0	0	50	6	1	0
12 Hawaii	17,638	141	22 b	11	0	3	0
13 Idaho	19,863	117	N/A	8	1	0	0
14 Illinois	181,984	1,388	0	60	N/A	15	2
15 Indiana	87,891	3,057	269	26	0 DA	3	0 DA
16 Iowa	38,418	282	10	25	0	0	0
17 Kansas	39,232	1,537	183	33	ND	1	1
18 Kentucky	54,423	335	N/A	20	0	0	0
19 Louisiana	68,275	332	4	10	DA	DA	DA
20 Maine	13,462	117	0	9	0	0	0
21 Maryland	69,574	811	101 e	15	1	0	49 f
22 Massachusetts	82,673	1,274	0	38	1	1	0
23 Michigan	134,889	6,401	106	34	2	6	63
24 Minnesota	67,546	392	0	22	4	5	0
25 Mississippi	42,980	30	0	2	0	0	0
26 Missouri	78,302	522	0	34	1	3	0
27 Montana	10,927	88	33	2	0	0	1
28 Nebraska	24,961	114	2	8	DA	DA	DA
29 Nevada	30,387	712	0	8	0	0	0
30 New Hampshire	13,987	75	0	16	0	0	0
31 New Jersey	112,311	2,518	0	60	7	54	23
32 New Mexico	26,809	656	NA	16	N/C	N/C	NC
33 New York	259,995	4,569	NC	151	4	NA	22
34 North Carolina	121,347	2,877	N/D	30	DA	DA	DA
35 North Dakota	8,847	116	3	3	0	3	0
36 Ohio	155,943	2,492	0	75	1	3	1
37 Oklahoma	48,650	321	14	24	0	8	0
38 Oregon	46,790	870	0	26	0	0	0
39 Pennsylvania	146,857	0	0	46	0	0	0
40 Rhode Island	13,180	0	0	4	0	2	0
41 South Carolina	53,562	3,016	284	23	0	11	13
42 South Dakota	10,589	75	0 N/D	5	0 N/D	0 N/D	0 N/D
43 Tennessee	84,832	448	3	59	N/C	N/C	N/C
44 Texas	368,019	5,022	142	157	0	27	4
45 Utah	48,454	292	9	23	3	17	23
46 Vermont	6,277	70	0	3	0	64	0
47 Virginia	96,755	1,619	0	18	0	9	1 c
48 Washington	80,453	413	3	20	N/A	2	N/A
49 West Virginia	21,620	330	0	6	0	0	0
50 Wisconsin	68,250	253	6	24	0	3	0
51 Wyoming	5,847	46	2	2	0	0	0
52 Puerto Rico	59,461	1,102	253	6	0	75	0
53 Virgin Islands	1,851	13	0	0	0	0	0
TOTAL	4,125,135	48,952 d	1,535 d	1,635 d	43 d	362 d	229 d

a = TBG; b = expired; c = hypopituitarism; d = total given does not include all states; e = 76 babies were premature or very sick and expired;

f = these were premature babies, placed on treatment for a short period generally challenged before discharge.

Table 4.15: Summation Results of Testing for Hypothyroidism

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screening calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary 2nd Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Alabama	62,562	62,173	99.38%			62,731	* 100.27%	420	0.67%
2 Alaska	9,866	9,821	99.54%			7,759	78.64%	313	3.17%
3 Arizona	85,470	81,956	95.89%			57,352	67.10%		
4 Arkansas	36,840	36,456	98.96%					567	1.54%
5 California	532,610	527,297	99.00%					3,392	0.64%
6 Colorado	65,679	63,219	96.25%	60,530	92.16%				
7 Connecticut	43,370	43,728	* 100.83%					290	0.67%
8 Delaware	11,639	11,714	* 100.64%	10,692	91.86%				
9 District of Columbia	15,159	15,125	99.78%					287	1.89%
10 Florida	204,305	204,030	99.87%						
11 Georgia	133,524	189,498	* 141.92%						
12 Hawaii	17,638	17,612	99.85%					247	1.40%
13 Idaho	19,863	20,254	* 101.97%			13,804	69.50%	202	1.02%
14 Illinois	181,984	188,164	* 103.40%						
15 Indiana	87,891	87,639	99.71%					31,782	36.16%
16 Iowa	38,418	38,141	99.28%					1,439	3.75%
17 Kansas	39,232	39,031	99.49%						
18 Kentucky	54,423	54,515	* 100.17%						
19 Louisiana	68,275	67,843	99.37%						
20 Maine	13,462	13,341	99.10%						
21 Maryland	69,574	72,390	* 104.05%	61,150	87.89%				
22 Massachusetts	82,673	82,703	* 100.04%					8,818	10.67%
23 Michigan	134,889	134,022	99.36%						
24 Minnesota	67,546	68,402	* 101.27%					887	1.31%
25 Mississippi	42,980	44,075	* 102.55%						
26 Missouri	78,302	78,640	* 100.43%			13,295	16.98%	5,282	6.75%
27 Montana	10,927	10,825	99.07%			3,543	32.42%		
28 Nebraska	24,961	24,863	99.61%						
29 Nevada	30,387	30,659	* 100.90%	25,668	84.47%				
30 New Hampshire	13,987	13,879	99.23%			815	5.83%		
31 New Jersey	112,311	112,241	99.94%					10,716	9.54%
32 New Mexico	26,809	25,745	96.03%	23,581	87.96%			168	0.63%
33 New York	259,995	258,449	99.41%						
34 North Carolina	121,347	120,750	99.51%						
35 North Dakota	8,847	8,806	99.54%					494	5.58%
36 Ohio	155,943	160,566	* 102.96%					37,021	23.74%
37 Oklahoma	48,650	52,760	* 108.45%						
38 Oregon	46,790	46,879	* 100.19%	43,423	92.80%			874	1.87%
39 Pennsylvania	146,857	145,874	99.33%						
40 Rhode Island	13,180	13,150	99.77%					427	3.24%
41 South Carolina	53,562	52,176	97.41%					3,657	6.83%
42 South Dakota	10,589	10,582	99.93%			180	1.70%	51	0.48%
43 Tennessee	84,832	79,539	93.76%						
44 Texas	368,019	355,100	96.49%	348,132	94.60%			1,919	0.52%
45 Utah	48,454	47,423	97.87%	46,460	95.88%			571	1.18%
46 Vermont	6,277	6,040	96.22%					326	5.19%
47 Virginia	96,755	99,410	* 102.74%					12,708	13.13%
48 Washington	80,453	76,886	95.57%			69,187	86.00%	1,551	1.93%
49 West Virginia	21,620	21,321	98.62%						
50 Wisconsin	68,250	66,614	97.60%			1,196	1.75%	1,284	1.88%
51 Wyoming	5,847	5,480	93.72%						
52 Puerto Rico	59,461	57,155	96.12%					843	1.42%
53 Virgin Islands	1,851	1,851	100.00%			10	0.54%	20	1.08%
TOTAL	a 4,125,135	4,156,812	b	619,636	b	229,872	b	126,752	b

* Percentage > 100% denotes inability of program to separate infants screened from specimens received; a = totals should be viewed as a rough estimate due to inability.

of many states, to know actual number of infants screened. Many programs count sample received but cannot eliminate duplication when multiple samples are received.

b = cannot be calculated due to inaccurate data.

5. Galactosemia

5.0 Introduction

Screening for galactosemia began in some screening programs in the 1960's although most programs developed their programs later. In some programs, there is still debate over the value of screening given that not all detected patients will experience optimal outcome. Most programs use a fluorescent testing procedure to screen for transferase deficient newborns (classical galactosemia), although some use protocols that include other microbiological techniques, including testing for total galactose in an attempt to identify other types of galactosemia (kinase and epimerase). Quick specimen analysis and result turnaround are considered essential elements of a screening program for galactosemia since devastating effects can be realized within a few days after birth. All data reported here was collected from questionnaires submitted to programs as noted in Chapter 3.

5.1 Definitions for Classical Galactosemia

Programs were asked to give the definition used by their program to define classical galactosemia. There is still not general agreement among programs as to the appropriate definition to be used for classical galactosemia. It is hoped that eventually programs can come to a consensus as to the best definition to use and the extent to which a metabolic specialist should be involved in the final diagnosis. From the definitions submitted, it is the usual case that a specialist is consulted to make the final diagnosis and program guidance is limited.

5.2 Definitions for Galact-osemia Variant

In addition to detecting classical galactosemia, many programs also detect other variant forms. Rather than offer a definition, the questionnaire asked programs to give their definition(s) for

variant forms of galactosemia for comparison with others. Table 5.02 gives the results of this survey.

5.3 Laboratory Techniques

While most U.S. programs began galactosemia screening using the Beutler fluorescence test, other techniques are available and are used in some programs. In order to different laboratory techniques in use, programs were asked to report their laboratory protocols. These data are reported in Table 5.03.

5.4 Initial Screening Results

In order to ascertain the effectiveness of screening for galactosemia, programs were asked to report their findings on initial screening. Initial screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of newborns screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated. Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 5.04. Programs wishing to explain some of their responses included information as footnotes to the table. Because programs continue to report inaccurate or inappropriate data in some of their responses, no totals were given in some columns of the table. Care should be taken in using these data, since not all programs reported data.

5.5 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when the second test is performed without regard for the initial screening result. Programs who requested a second screen because of either a certain result on

the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as ‘repeat screens.’

For each case, the data requested included the total newborns tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. It should be recognized that this is a different way of reporting these data than previously and may, therefore, be somewhat confusing. It is hoped that these data better represent the findings from additional screens. Once again, the reader is cautioned about using the data without regard to notes of explanation.

5.6 Cases of Classical Galactosemia Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of classical galactosemia, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 5.06. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

5.7 Cases of Variant Forms of Galactosemia Divided by Sex and Race/Ethnicity

Programs were also asked to report all diagnosed cases of variant forms of galactosemia that might have been detected (if these data are kept) and these data are reported in Table 5.07.

5.8 Time Until Treatment

In order to look at overall program efficiency, programs were asked to report the number of days from birth until treatment of classical galactosemia. The general definition of treatment used in the questionnaire was “initiation of dietary or drug regimen” and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were

asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 5.08.

5.9 Historic Data

In order to document the value of screening for galactosemia, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of various types of cases detected over the years. These data are given in Table 5.09. Since many programs may not have reported these data, the reader is cautioned about using them out of context.

5.10 Total Newborns Screened

Table 5.10 gives a summation of the data contained in Tables 5.04 and 5.05 along with the number of births in each state or territory as reported in Table 1.01. This table was included to provide a tabulation of these data without having to refer to several tables.

5.11 Summation of Results

Table 5.11 sums the significant testing data from this Chapter. It includes the total births from Table 1.01, the total newborns reported as being screened, and the testing data reported in Tables 5.04 and 5.05. Comparative percentages have been provided at the request of a number of readers, but in many cases these data are based on estimates or on data that appear to contain duplicate information, thus total compliance percentages may exceed 100% in some cases.

Table 5.01: Definitions for Classical Galactosemia

State/Territory	Definition for Classical Galactosemia
1 Alabama	<2.4 U/gHb.
2 Alaska	Phenotype GG and enzyme activity < 1.0 units.
3 Arizona	Lack of enzyme activity.
4 Arkansas	Determined by metabolic specialist; complete absence of GALT activity on whole blood assay.
5 California	Genotype GG based on banding and parent studies.
6 Colorado	Zero activity of RBC Galactose-1-P-uridyltransferase.
7 Connecticut	Classified by treatment center.
8 Delaware	Deficient uridyl transferase activity & elev. total Gal. Diagnostic/classification made by med. consultant; supported by diagnostic tests.
9 District of Columbia	Deficient uridyltransferase activity and elevated total galactose.
10 Florida	Absence of red cell Galactose-1-Phosphate-uridyl-transferase.
11 Georgia	GALT < 1level of activity by quantitative enzyme analysis. Confirmed by DNA & Isozyme analysis.
12 Hawaii	Phenotype GG and enzyme activity <1.0 units.
13 Idaho	Phenotype GG and enzyme activity < 1.0 units.
14 Illinois	Diagnosis and classification determined by pediatric medical consultant.
15 Indiana	Deficient transferase activity with increased total galactose. Confirmed with WB GAL-1-PUT and genotyping.
16 Iowa	Deficient Gal-1-P uridyl transferase activity, confirm by electrophoresis and/or DNA.
17 Kansas	Galactose-1-phosphate uridyl transferase has no activity.
18 Kentucky	G/S = GALT level approx. 0.0 in mol/hr/gm; G-1-P levels generally elevated-most levels >15 mL%, EP shows 2 gal alleles.
19 Maine	Diagnosis & classification made by medical consultant and/or reference lab performing the diagnostic tests.
20 Maryland	Absent transferase, elevated Gal/Gal-1-P.
21 Massachusetts	Diagnosis and classification determined by metabolic consultant.
22 Michigan	Galactose metabolism disorder due to a deficiency in the enzyme GALT. Designated as a GG genotype.
23 Minnesota	No or very low GALT activity, total Gal (Gal & Gal-1-P) ≥ 20 mg/dL. Classification determined by metabolic specialist.
24 Mississippi	Defined by genetic center; generally absent enzyme, G/G, or other DNA mutation.
25 Missouri	Diagnosis and classification determined by medical consultant.
26 Montana	Enzyme activity of < 3.1 u/gHB and total metabolite of > 12.6 mg/dL (total galactose).
27 Nebraska	GG genotype; transferase activity very low or absent; classification confirmed by metabolism specialist.
28 Nevada	Abnormal Beutler + blood glucose > 10 mg/dL; Abnormal Beutler + galactose < 10 mg/dL.
29 New Hampshire	Diagnosis and classification made by metabolic consultant.
30 New Jersey	Diagnosis and classification of G/G confirmed by medical specialist.
31 New Mexico	Genotype classification defined by banding pattern at confirmatory lab.
32 New York	Diagnosis and classification determined by treatment center.
33 North Carolina	Diagnosis and classification made by medical consultant.
34 North Dakota	Diagnosis confirmed by genotyping and enzymology testing.
35 Ohio	Diagnosis and classification made by medical consultant.
36 Oklahoma	GAL-1-PUT < 5 u/g; GG genotype
37 Oregon	Phenotype GG and enzyme activity < 1.0 units.
38 Pennsylvania	NR
39 Rhode Island	Diagnosis and classification determined by metabolic consultant.
40 South Carolina	Determined by specialist.
41 South Dakota	Diagnosis and classification is determined by medical consultant.
42 Tennessee	Defined by genetic centers: generally absent enzyme G/G or other DNA mutations.
43 Texas	Very low or no activity, galactose-1-phosphate uridyltransferase.
44 Utah	GAL-1-P uridyltransferase genotype of GG.
45 Vermont	Enzyme activity absent; gal > 20 mg/dL, gal-1-P ≥ 50 mg/dL.
46 Virginia	Demonstration of uridyl transferase deficiency with elevated total galactose levels.
47 West Virginia	GAL-1-Phosphate uridyl transferase activity deficient with elevated total galactose levels of ≥ 20mg/dL.
48 Wisconsin	Diagnosis and classification made by treatment center.
49 Wyoming	Zero activity of RBC Galactose-1-P-uridyltransferase.
50 Puerto Rico	Galactosemia caused by deficiency of Galactose-1-Phosphate uridyl transferase.
51 Virgin Islands	Zero Enzyme activity of transferase enzyme.
52 NeoGen Screening	Deficient uridyltransferase activity and elevated total galactose.

Table 5.02: Definitions for Galactosemia Variant

State/Territory	Definition for Galactosemia Variant
1 Alabama	2.5 - 4.0 U/gHb.
2 Alaska	Phenotype other than GG but not G/N.
3 Arizona	Reduced enzyme activity. (Arizona does not actively screen for variants).
4 Arkansas	Determined by metabolic specialist; < 30% GALT activity on whole blood assay; or confirmed epimerase or galactokinase deficiency.
5 California	DG only.
6 Colorado	NC
7 Connecticut	Classified by treatment center.
8 Delaware	Deficient uridyl transferase activity. Diagnostic & classification made by medical consultant; supported by diagnostic tests.
9 District of Columbia	Galactose deficiency; galto epimerase deficiency, or Duarte/Galactosemia compound heterozygote.
10 Florida	> 0% or < 25% red cell GALT activity; may have genotyping performed; D/G; double heterozygote/approximately 50% red cell GALT activity; N/G carrier.
11 Georgia	GALT activity ~ 25% by quantitative enzyme analysis. Confirmed by Isozyme & DNA analysis.
12 Hawaii	Phenotype other than GG but not G/N.
13 Idaho	Phenotype other than GG but not G/N.
14 Illinois	Diagnosis and classification determined by pediatric medical consultant.
15 Indiana	Deficient transferase activity with abnormal total galactose. Confirmed with WB GAL-1-PUT and genotyping.
16 Iowa	Reduced activity of Gal-1-P uridyl transferase activity. Confirmation by electrophoresis and/or DNA.
17 Kansas	Transferase activity > 50% but less than 100%.
18 Kentucky	Double heterozygous both the Duarte allele & classical allele on EP. 25% normal GALT (4-7), G-1-P may or may not be elev.
19 Maine	Diagnosis & classification made by medical consultant and/or reference lab performing the diagnostic tests.
20 Maryland	Decreased transferase activity; elevated Galactose/Gal-1-P; electrophoresis and DNA to establish genotype.
21 Massachusetts	Diagnosis and classification determined by metabolic consultant.
22 Michigan	Partial GALT enzyme activity (DG, DD, LAG), deficiency in other GAL enzyme (epimerase or galactokinase deficiency).
23 Minnesota	Partial GALT activity and total Gal < 20 mg/dL. Classification determined by metabolic specialist.
24 Mississippi	Defined by genetic centers; generally low enzyme, D/G, or other DNA mutation.
25 Missouri	Diagnosis and classification determined by medical consultant.
26 Montana	Enzyme activity of slightly less than 3.1 u/gHb and total metabolite of < 12.6 mg/dL total galactose, confirm by serum tests and DNA.
27 Nebraska	DG genotype or other mixed heterozygote; transferase activity < 50%; classification confirmed by metabolism specialist.
28 Nevada	Normal Beutler + galactose > 10 mg/dL.
29 New Hampshire	Diagnosis and classification made by metabolic consultant.
30 New Jersey	Decreased transferase activity or other genotype which is not normal, confirmed by medical specialist.
31 New Mexico	Genotype classification defined by banding pattern at confirmatory lab.
32 New York	Diagnosis and classification made by treatment center.
33 North Carolina	Diagnosis and classification made by medical consultant.
34 North Dakota	Diagnosis confirmed by genotyping and enzymology testing.
35 Ohio	Diagnosis and classification made by medical consultant.
36 Oklahoma	GAL-1-PUT < 10 u/g; DG genotype, Duarte or NG carrier genotype.
37 Oregon	Phenotype other than GG but not G/N.
38 Pennsylvania	NR
39 Rhode Island	Diagnosis and classification determined by metabolic consultant.
40 South Carolina	Determined by specialist.
41 South Dakota	Diagnosis and classification is determined by medical consultant.
42 Tennessee	Defined by genetic centers: generally low enzyme, D/G or other DNA mutation.
43 Texas	Diagnosed by consultant.
44 Utah	GAL-1-P uridyl transferase genotype other than normal or GG.
45 Vermont	Low enzyme activity; gal ≤ 14 mg/dL, gal-1-P ≥ 50 mg/dL.
46 Virginia	Heterozygous galactosemia: duarte variant demonstrating uridyl transferase deficiency.
47 West Virginia	Gal-1-PUT activity < 50% with total galactose ≥ 10 mg/dL ≤ 20 mg/dL confirmed by genotyping.
48 Wisconsin	Diagnosis and classification made by treatment center.
49 Wyoming	Gal-1-Put activity <50% & >5%, most have Duarte on EP; prog. detects galactokinase & epimerase deficiency, some carriers class. gal.
50 Puerto Rico	Confirmed case of galactosemia with repeat normal result of galctose-1-phosphate uridyl transferase.
51 Virgin Islands	Diagnosis determined by genotyping.
52 NeoGen Screening	Galactokinase deficiency, galactoeipimerase deficiency, or Duarte/Galactosemia compound heterozygote.

Table 5.03: Galactosemia - Laboratory Testing

State/Territory	Buetler (B) (TRANS-FERASE)	Microbiological Inhibition Assay (M) (Galactose)	Other (O)	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Immediate Serum Follow-up</i>
1 Alabama			Quantitative Fluorometric	< 4.0 U/gHb.	< 2.4 U/gHb.
2 Alaska	X	X	Hill Test	Beutler NF, ≥ 72 hrs. old or Beutler normal, galactose = 20-29 mg/dL, galactose > 30 mg/dL Beutler normal.	Beutler NF, gal > 20mg/dL; Beutler NF < 72 hrs old or not on lactose formula, feeding history unknown or transfused.
3 Arizona			Fluorometric for GALT	≤ 2.4 U/gHb on 1st and 2nd screen with no clin. symptoms.	< 2.4 U/gHb on 1st screen with clinical symptoms.
4 Arkansas			Fluorometric	Total Gal 10-15 mg/dL; GALT 3.6-5.0 U/gHb Gal-1-P ≥ 4.0 or < 15 gal, ≤ 3.5 GALT, and G-1-P, unacceptable S1A (inconclusive/Partial)	Any gal < 3.5 GALT; any Gal-1-P or > 15 gal; any GALT; any Gal-1-P (Postive)
5 California	X			NC	50 mg/dL
6 Colorado	X			Little or no fluorescence.	Little or no fluorescence.
7 Connecticut			X	NA	All abnormal require immediate serum follow-up.
8 Delaware	Colorimeter		Neometrics-accuwell	GALT < 1.4 U/gm Hgb	GALT < 2.3 U/gm Hgb
9 District of Columbia	X		Fluorescent Assay	Elevated Total GAL, normal uridylyltransferase	Elevated significant Total GAL, deficient uridylyltransferase.
10 Florida			X	2.5-3.4 repeat test same sample, if still 2.5-3.4 repeat	< 2.4 with SIA > 18.2
11 Georgia	X	X		Inconclusive result (reduced but not absent fluorescence) no more than slight elev. of gal (normal ≤ 16).	Pos. screen (no fluorescence) with elevated galactose > 15 (normal < 10)
12 Hawaii	X	Back-up only	Hill test - fluorometric	Abnormal Beutler and < 20 mg/dL Gal.	Abnormal Beutler and > 20 mg/dL Gal.
13 Idaho	X	Back-up only	Fluorometric, manual (galactose) methodology.	Beutler no fluor. > 72 hrs. old or Beutler normal, galactose 15-29 mg%; Galactose > 30 mg%, Beutler normal.	Gal > 15mg%, or Beutler, no Fluor. screened, < 72 hrs. old or feeding history unknown or transfused.
14 Illinois	Secondary		Tot Gal - Fluorometric	≥ 7 mg/dL	≥ 7 mg/dL Beutler absent or trace
15 Indiana	X			GALT 1.5 - 3.0 Ugm/Hgb	GALT < 1.5 Ugm/Hgb
16 Iowa			Isolab - EIA	> 3.1 ≤ 3.7 mg/dL	≤ 3.1 mg/dL
17 Kansas	X			None (No secondary F/U)	None
18 Kentucky	X			1st time heterozygous spec. receive further testing.	Positive controls & heterozygous on 2 successive spec.
19 Maine			Fluorometric Assay	≥ 14 mg/dL - reduced or absent GALT	
20 Maryland	X		Hill (Gal + Gal-1-P)	B=normal, Hill Gal > 10 mg/dL but < 40 mg/dL; B=abnormal, Hill ≤ 10 mg/dL.	B=abnormal, Hill > 10 mg/dL; B = normal, Hill > 40 mg/dL.
21 Massachusetts			Fluorometric total GAL	if ≥ 6 mg/dL then do Beutler test	if ≥ 10 mg/dL retest tot gal and do Beutler; ≥ 14 retest
22 Michigan			Fluorometric for GALT	GALT 2.5 - 3.0 U/gHb	GALT ≤ 2.4 U/gHb
23 Minnesota	X		T Gal in-house	≥ 10 mg/dL	≥ 20 mg/dL
24 Mississippi	X		Fluorimetric-Alpkem.	Total Gal > 7 mg/dL < 15 mg/dL.	Gal > 15mg/dL; Enzyme Absent. low, normal, absent.
25 Missouri	X		Gal and GAL-1-P	Abnormal Beutler and < 10 mg/dL Gal.	Abnormal Beutler and ≥ 10 mg/dL Gal.

continued

Table 5.03: Galactosemia - Laboratory Testing (continued)

State/Territory	Buetler (B) (TRANS-FERASE)	Microbiological Inhibition Assay (M) (Galactose)	Other (O)	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Immediate Serum Follow-up</i>
26 Montana			Fluorometric	< 3.1 & 12-20 mg/dL; Hill - 14-20 mg/dL	< 3.1 u/gHb & > 20 mg/dL; Hill - > 20 mg/dL
27 Nebraska	X				Less fluorescence than known normal control
28 Nevada	X		Hill Test	> 10 mg/dL	> 30 mg/dL
29 New Hampshire			Total GAL - if abn. then Beutler	If ≥ 6 mg/dL then do Beutler; If >10 retest Tot. GAL & do Beutler.	Phone follow-up when: Total GAL ≥ 20 mg/dL & enz. reduced. Total GAL >30 or enzyme absent; if >14 mg/dL rep. req.
30 New Jersey			FIA (transferase) / FIA (GAL)	> 3.1-3.5 units/gHb / ≥ 7.2 mg/dL	≤ 3.1 units/gHb / DA
31 New Mexico	X		Modified Hill		Positive - no fluorescence and ≥ 10 µg/dL
32 New York	X		Modified Hill	DA	Beutler = no fluorescence and Hill ≥ 10 mg/dL.
33 North Carolina			Fluorometric, total gal.	Total GAL ≥ 10 mg/dL no UT activity.	≥ 10 mg/dL
34 North Dakota			Isolab EIA	≥ 3.1; ≤ 4.0	≤ 3.1
35 Ohio	X				No Fluorescence.
36 Oklahoma	Supplemental		Total GAL	≥ 10 mg/dL enzyme present; > 7 mg/dL enzyme low	≥ 7 mg/dL enzyme absent
37 Oregon	X		Fluorometric	Pos Beutler > 72 hrs old or Beutler normal, Gal > 20 mg%.	Pos Beutler, Gal ≥ 20 mg% or Pos Beutler < 72 hrs old or not on Lactose or feeding history not known or transfused.
38 Pennsylvania			Astoria Pacific Spotcheck		
39 Rhode Island			Tot. GAL by bact. phage assay	14-29.9 with 23 enzymes; < 20 with 1 enzyme	20-29.9 with 1 enzyme; ≥ 30 or 0 enzyme.
40 South Carolina	X a	DA	Microfluorometric	Total galactose ≥ 10 mg/dL and/or transferase enzyme deficient.	DA
41 South Dakota	X				
42 Tennessee			Fluorometric - RFA	≥ 7 mg/dL enzyme low, ≥15 - <20 mg/dL enzyme normal.	≥15 mg/dL enzyme low, ≥20 enzyme normal
43 Texas		X		≥ 15 mg/dL	2 abnormal screens or 1 very elevated screen
44 Utah			Fluorometric enzyme assay	≤ 4 mg/dL	< 4 mg/dL
45 Vermont			G-1-P abnormal	Gal 14 - 29.9 mg/dL (age dependent)	Gal. ≥ 30 mg/dL or GALT absent.
46 Virginia	X		Hill fluorometric tot. gal method.	< 50% fluorescence on Beutler and/or Hill ≥ 10 mg/dL.	Hill ≥ 15 mg/dL or 3 consecutive sample w/abnormal Beutlers.
47 West Virginia	X		Fluorometric Total Gal.	GALT < 50% + total galactose = ≥ 10 mg/dL ≤ 20 mg/dL	Gal-1-PUT deficient or total gal ≥ 20 mg/dL.
48 Wisconsin	X b		Fluorometric, Total gal.	≥ 13 mg/dL total gal or no qualitative Gal-1-Put	
49 Wyoming	X			Little or no fluorescence.	Little or no fluorescence.
50 Puerto Rico	X		Hill Test	> 10 mg/dL	
51 Virgin Islands	X			No fluorescence	Little or no fluorescence.
52 NeoGen Screening	X		GAL Only cont. flow assay	Total Gal. ≥ 15 mg/dL < critical cutoff; UT wh1	uridyltransferase ≤ 60 µM

a = Beutler test done only if the total galactose is ≥ 10 mg/dL or infant is receiving a non-lactose feeding, added enzyme test for all infants whose specimen is collected before 24 hours of age in April 1996;

b = performed on highest 3% of daily total galactose samples.

Table 5.04: Initial Screening Results - Galactosemia

State/Territory	Number of Newborns Screened	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number of Newborns Confirmed w/ Classical Galactosemia	Number of Newborns Confirmed w/Variant Galactosemia
1 Alabama	61,894	NC		1	
2 Alaska	9,821	3			1
3 Arizona	81,956	11		2	2
4 Arkansas	36,702	64	3		19
5 California	527,297	143		5	32
6 Colorado	63,219	8			NC
7 Connecticut	43,722	21		1	7
8 Delaware	11,714	4	NA		4
9 District of Columbia	15,125	45	25 a		3
10 Florida	204,030	14		7	
11 Georgia	189,498			5	18
12 Hawaii	17,612	18	7 b		3
13 Idaho	20,254	10			
14 Illinois	188,164 g	115	3	2	8
15 Indiana	87,639	117	8	2	52
16 Iowa	38,141	30	1	1	22
17 Kansas	39,031	5		1	
18 Kentucky	54,515	84			5
19 Maine	13,341	12			
20 Maryland	72,390	585	2		
21 Massachusetts	82,703	158	N/A		
22 Michigan	134,022	139		6	
23 Minnesota	68,402	7		2	5
24 Mississippi	44,075	34			
25 Missouri	78,640	650		1	100
26 Montana	10,825	2			
27 Nebraska	24,863	12		1	3
28 Nevada	30,659	5		1	3
29 New Hampshire	13,879	7			
30 New Jersey	112,241	166		1	33
31 New Mexico	25,745	33			14
32 New York	258,449	9	N/D	4	2
33 North Carolina	120,750	10	2		5
34 North Dakota	8,806	11			7
35 Ohio	160,566	152		3	122
36 Oklahoma	52,760	238	7		10
37 Oregon	46,879				3
38 Pennsylvania	36,326	67			11
39 Rhode Island	13,150				
40 South Carolina	52,176	325	39 d	1	13
41 South Dakota	10,220	9		1	6
42 Tennessee	79,539	165	10	3	23
43 Texas	355,100			2	19
44 Utah	47,423	55			41
45 Vermont	6,040	4			
46 Virginia	99,410	211		2	11
47 West Virginia	21,321	79		1	
48 Wisconsin	66,614	110		3	10
49 Wyoming	5,480				
50 Puerto Rico	57,048	22			
51 Virgin Islands	1,851	14			
52 NeoGen Screening	140,789	122	18 c	1	26
TOTAL	e	e	e	60 f	643 f

a = lab lost to follow-up - DC MCH has more info; b = 6 expired, 1 refused; c = physician notification/baby ctb;

d = none with deficient enzyme; e = totals not given - too many programs reported inaccurate or inappropriate data; f = total given does not include all states; g = includes repeat specimens.

Table 5.05: Second Screens for Galactosemia

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed w/Classical Galactosemia				Number of Newborns Confirmed w/Variant Galactosemia			
	R	D	P		R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total
1 Alabama		N/C	1,946	1,946																
2 Alaska		7,759	215	7,974	5		5													
3 Arizona		57,352		57,352	17		17								5				5	
4 Arkansas			599	599			11	11												
5 California			3,392	3,392																
6 Colorado	60,530		N/C	60,530	11		N/C													
7 Connecticut			219	219			8	8												
8 Delaware	10,692			10,692					N/A		N/A	N/A								
9 District of Columbia			287	287																
10 Florida																				
11 Georgia		ND	ND																	
12 Hawaii			197	197																
13 Idaho		13,804	199	14,003	8		8													
14 Illinois			N/C																	
15 Indiana		DA	31,782	31,782			251	251			5	5						36	36	
16 Iowa		DA	1,439	1,439		DA				DA				DA						
17 Kansas																				
18 Kentucky	NR																			
19 Maine																				
20 Maryland	61,152			61,152	55		55	10			10				7				7	
21 Massachusetts			8,818	8,818			12	12												
22 Michigan															13				13	
23 Minnesota			319	319			24	24												
24 Mississippi																				
25 Missouri		13,295	5,282	18,577																

continued

Table 5.05: Second Screens for Galactosemia (continued)

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed w/Classical Galactosemia				Number of Newborns Confirmed w/Variant Galactosemia			
	R	D	P		R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total
26 Montana		3,540		3,540																
27 Nebraska		DA	DA																	
28 Nevada	25,668			25,668	31			31												
29 New Hampshire		815		815																
30 New Jersey		DA	10,716	10,716		DA	39	39												
31 New Mexico	953		84	1,037	21		4	25												
32 New York		DA				DA	N/C			DA	N/C		DA	N/C			DA	N/C		
33 North Carolina		N/C	b	N/C																
34 North Dakota		DA	494	494		DA				DA			DA				DA			
35 Ohio			37,021	37,021																
36 Oklahoma		N/C	N/C			N/C	N/C			N/C	N/C		N/C	N/C			N/C	N/C		
37 Oregon	43,423		862	44,285																
38 Pennsylvania																				
39 Rhode Island			427	427																
40 South Carolina		N/A	3,657	3,657		N/A				N/A			N/A				N/A			
41 South Dakota		139	42	181			6	6												
42 Tennessee																				
43 Texas	348,132		1,919	350,051						1			1		13				13	
44 Utah	18,883 a		406	19,289	65			65	1			1			61				61	
45 Vermont			326	326																
46 Virginia			12,594	12,594			173	173												
47 West Virginia			N/A			5	48	53												
48 Wisconsin		1,196	1,230	2,426			17	17												
49 Wyoming	DA	DA	N/C		DA	DA	N/C		DA	DA	N/C		DA	DA	N/C		DA	DA	N/C	
50 Puerto Rico			22	22																
51 Virgin Islands			13	13																
52 NeoGen Screening		1,984	1,546	3,530 c																
Total	d	569,433	99,884	126,053	e			e				e	1		1	f	81	18	36	135 f

a = unable to distinguish between 1st & 2nd screens for 6 mo. due to switch of computer software; **b** = number of specimens rather than number of infants were counted; **c** = these numbers are overall; **d** = totals given reflect program responses and should be viewed as estimates only, given the number not reporting and the number of caveats listed; **e** = totals not given - too many programs reported inaccurate or inappropriate data; **f** = total given does not include all states.

Table 5.06: Cases of Classical Galactosemia

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Alabama		1												1	1		
2 Alaska																		
3 Arizona		2												2	2			
4 Arkansas																		
5 California	1	1			1		1						3	1	5 ³	1		1
6 Colorado																		
7 Connecticut	1												1		1	1		1
8 Delaware																		
9 District of Columbia																		
10 Florida	3	2	1								1		5	2	7			
11 Georgia		3	1	1									1	4	5			
12 Hawaii																		
13 Idaho																		
14 Illinois	2												2		2			
15 Indiana		2												2	2			
16 Iowa		1												1	1	1		1
17 Kansas		1												1	1			
18 Kentucky																		
19 Maine																		
20 Maryland																		
21 Massachusetts																		
22 Michigan		6												6	6	1		1
23 Minnesota	1	1											1	1	2			
24 Mississippi																		
25 Missouri	1													1	1			
26 Montana																		
27 Nebraska		1												1	1			
28 Nevada		1												1	1			
29 New Hampshire																		
30 New Jersey			1											1	1			
31 New Mexico																		
32 New York	1			1					1				1	2	4 ³	1		1
33 North Carolina																		
34 North Dakota																		
35 Ohio											3		3	3				
36 Oklahoma																		
37 Oregon																		
38 Pennsylvania																		
39 Rhode Island																		
40 South Carolina	1													1	1			
41 South Dakota											1		1	1				
42 Tennessee	1	2											1	2	3			
43 Texas				1										1	3 ³	2		2
44 Utah																		
45 Vermont																		
46 Virginia			1	1										1	1	2		
47 West Virginia		1												1	1			
48 Wisconsin	1	2												1	2	3		
49 Wyoming																		
50 Puerto Rico																		
51 Virgin Islands																		
52 NeoGen Screening		1												1	1			
TOTAL	13	28	4	4	1		1			1	5		24	33	61	1	6	7

¹did not report a breakdown of Race/Ethnicity.

²total reported is from Table 5.04 and Table 5.05; therefore total does not equal or may differ from data given in Table 5.06 and denotes a problem in reliability.

³Ethnicity counted as a Race and is included in total.

Table 5.07: Cases of Variant Galactosemia

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Alabama																	
2 Alaska											1	0	1	1				
3 Arizona	4	3											4	3	7	1	1	2
4 Arkansas	9	7	1	1									10	8	19 ³	1	0	1
5 California a	8	9	1								1	9	10	32 ³	8	5	13	
6 Colorado																		NC
7 Connecticut	3	3									1	3	4	7		2	2	
8 Delaware	3	1											3	1	4			
9 District of Columbia												3	0	3	3			
10 Florida																		
11 Georgia	5	10	1										6	10	18 ³		2	2
12 Hawaii					1	2							1	2	3			
13 Idaho																		
14 Illinois	2	2	1						1	1			4	3	8 ³	1		1
15 Indiana	40	44							1				41	44	88 ³		3	3
16 Iowa	1	5									7	9	8	14	22			
17 Kansas																		
18 Kentucky															5 ¹			
19 Maine																		
20 Maryland	1	1	1							1	2	1	4	3	7			
21 Massachusetts																		
22 Michigan	6	7											6	7	13	1	1	1
23 Minnesota	3	1			1								4	1	5			
24 Mississippi																		
25 Missouri	42	35	11	9		1					1	1	54	46	100	2		2
26 Montana																		
27 Nebraska															3 ¹			
28 Nevada		2											0	2	3 ³	1	1	1
29 New Hampshire																		
30 New Jersey	13	13		1							4	2	17	16	33			
31 New Mexico	1	8					2		2		1		6	8	14	3	4	7
32 New York	1												1	0	2 ³	1		1
33 North Carolina	1	4											1	4	5			
34 North Dakota	3	1					1	2					4	3	7			
35 Ohio											63	59	63	59	122			
36 Oklahoma	3	5					1	1					4	6	10			
37 Oregon	1	2											1	2	3			
38 Pennsylvania	2	5	2	1						1			4	7	11			
39 Rhode Island																		
40 South Carolina	6	3	2	2									8	5	13			
41 South Dakota												4	2	4	2	6		
42 Tennessee	13	7	3										16	7	23			
43 Texas	12	8	1	1					1	1			14	10	32 ³	3	5	8
44 Utah											55	47	55	47	102			
45 Vermont																		
46 Virginia	4	5							1				1	5	6	11		
47 West Virginia																		
48 Wisconsin	3	6		1									3	7	10			
49 Wyoming																		
50 Puerto Rico																		
51 Virgin Islands																		
52 NeoGen Screening	15	11											15	11	26			
TOTAL	205	208	24	16	2	3	4	3	6	4	137	128	378	362	778	20	24	44

¹ did not report a breakdown of Race/Ethnicity.

² total reported is from Table 5.04 and Table 5.05; therefore total does not equal or may differ from data given in Table 5.07 and denotes a problem in reliability.

³ Ethnicity counted as a Race and is included in total.

a = DG variants only.

Table 5.08: Days from Birth Until Treatment Initiated for Classical Galactosemia

Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	TOTAL
1 Alabama																1 ¹
2 Alaska																
3 Arizona			1				1									2
4 Arkansas																
5 California		2	1	1	1											5
6 Colorado																
7 Connecticut									1							1
8 Delaware																
9 District of Columbia																
10 Florida	4				1				1				1			7
11 Georgia	2					1		1					1			5
12 Hawaii																
13 Idaho																
14 Illinois														2		2
15 Indiana	1		1													2
16 Iowa				1												1
17 Kansas							1									1
18 Kentucky																
19 Maine																
20 Maryland																
21 Massachusetts																
22 Michigan	1	1	1	1	1	1										6
23 Minnesota		1			1											2
24 Mississippi																
25 Missouri					1											1
26 Montana																
27 Nebraska	1															1
28 Nevada													1			1
29 New Hampshire																
30 New Jersey					1											1
31 New Mexico																
32 New York			1	1	1		1									4
33 North Carolina																
34 North Dakota																
35 Ohio															3	3
36 Oklahoma																
37 Oregon																
38 Pennsylvania																
39 Rhode Island																
40 South Carolina	1															1
41 South Dakota															1	1
42 Tennessee															3	3
43 Texas							1						2			3
44 Utah																
45 Vermont																
46 Virginia	1	1														2
47 West Virginia											1					1
48 Wisconsin	1				2											3
49 Wyoming																
50 Puerto Rico																
51 Virgin Islands																
52 NeoGen Screening															1	1

¹did not report a breakdown of Days from Birth.

²total reported is from Table 5.04 and Table 5.05; therefore total does not equal or may differ from data given in Table 5.08 and denotes a problem in reliability.

Table 5.09: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Classical Galactosemia			Galactosemia Variant		
	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases
1 Alabama				N/C		N/C
2 Alaska	1987	13	2	1987	13	7
3 Arizona	1994	6	4	1994	6	20
4 Arkansas	1996	4	4	1996	4	65
5 California	1980	20	139	1980	20	407 a
6 Colorado	Mar-79	20 3/4	14	N/C		N/C
7 Connecticut	1964	36	25	1964	36	16
8 Delaware	1993	7	2	1993	7	17
9 District of Columbia	1996	4		1996	4	4
10 Florida	1978	22	66			
11 Georgia	Feb-79	20 1/9	62	Feb-79	20 1/9	265
12 Hawaii	Jul-97	2 1/2		Jul-97	2 1/2	5
14 Idaho	1976	24	9	1976	24	11
14 Illinois	1984	16	63	1984	16	50
15 Indiana	Jul-85	15 1/2	22	Jul-85	15 1/2	696
16 Iowa	N/C		N/C			
17 Kansas	1984	16	23			
18 Kentucky NR						
19 Maine	1976	24	13			
20 Maryland	1979	21	27	1993	7	75
21 Massachusetts	1964	36	42			
22 Michigan	1985	15	49	1987	13	250
23 Minnesota NR						
24 Mississippi	NR		NR	NR		NR
25 Missouri	NR		NR	NR		NR
26 Montana	1985	15	7	1993	7	4
27 Nebraska	Nov-96	4 1/6	1 b	NC		NC
28 Nevada NR						
29 New Hampshire	1983	17	5			
30 New Jersey NR						
31 New Mexico NR	1981	19	6	1981	19	107
32 New York	1968	32	140	1997	3	6
33 North Carolina NR						
34 North Dakota	1992	8		1992	8	22
35 Ohio NR						
36 Oklahoma	1991	9	10			
37 Oregon NR						
38 Pennsylvania						
39 Rhode Island NR						
40 South Carolina	Oct-92	8 1/4	7	Oct-92	8 1/4	56
41 South Dakota NR						
42 Tennessee NR						
43 Texas	1979	21	103	1979	21	107
44 Utah	1980	20	15	1980	20	510
45 Vermont	Jul-89	11 1/2		Jul-89	11 1/2	3
46 Virginia	1984	16	72	1984	16	N/A
47 West Virginia	N/D		N/D	N/D		N/D
48 Wisconsin	1978	22	26	1978	22	54
49 Wyoming	Apr-79	20 3/4	1	Apr-79	20 3/4	4
50 Puerto Rico	1998	2		1998	2	
51 Virgin Islands NR						
52 NeoGen Screening NR						

a = DG only; **b** = 1 case detected in 2000 & 1 case detected in 1995, but screening for Gal not started until 11/1/96.

Table 5.10: Total Newborns Screened for Galactosemia

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number of Newborns Confirmed w/ Classical Galactosemia	Number of Newborns Confirmed w/Variant Galactosemia
1 Alabama	62,562			1	
2 Alaska	9,866	8			1
3 Arizona	85,470	28		2	7
4 Arkansas	36,840	75	3		19
5 California	532,610	143		5	32
6 Colorado	65,679	19			NC
7 Connecticut	43,370	29		1	7
8 Delaware	11,639	4	NA		4
9 District of Columbia	15,159	45	25		3
10 Florida	204,305	14		7	
11 Georgia	133,524			5	18
12 Hawaii	17,638	18	7		3
13 Idaho	19,863	18			
14 Illinois	181,984	115	3	2	8
15 Indiana	87,891	368	13	2	88
16 Iowa	38,418	30	1	1	22
17 Kansas	39,232	5		1	
18 Kentucky	54,423	84			5
19 Maine	13,462	12			
20 Maryland	69,574	640	12		7
21 Massachusetts	82,673	170			
22 Michigan	134,889	139		6	13
23 Minnesota	67,546	31		2	5
24 Mississippi	42,980	34			
25 Missouri	78,302	650		1	100
26 Montana	10,927	2			
27 Nebraska	24,961	12		1	3
28 Nevada	30,387	36		1	3
29 New Hampshire	13,987	7			
30 New Jersey	112,311	205		1	33
31 New Mexico	26,809	58			14
32 New York	259,995	9	ND	4	2
33 North Carolina	121,347	10	2		5
34 North Dakota	8,847	11			7
35 Ohio	155,943	152		3	122
36 Oklahoma	48,650	238	7		10
37 Oregon	46,790				3
38 Pennsylvania	146,857	67			11
39 Rhode Island	13,180				
40 South Carolina	53,562	325	39	1	13
41 South Dakota	10,589	15		1	6
42 Tennessee	84,832	165	10	3	23
43 Texas	368,019			3	32
44 Utah	48,454	120	1		102
45 Vermont	6,277	4			
46 Virginia	96,755	384		2	11
47 West Virginia	21,620	132		1	
48 Wisconsin	68,250	127		3	10
49 Wyoming	5,847				
50 Puerto Rico	59,461	22			
51 Virgin Islands	1,851	14			
52 NeoGen Screening	144,319	122	18	1	26
TOTAL	4,120,726	4,916 a	141 a	61 a	778 a

a = total given does not include all states.

Table 5.11: Summation Results of Testing for Galactosemia

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screen calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary 2nd Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Alabama	62,562	61,894	98.93%					1,946	3.11%
2 Alaska	9,866	9,821	99.54%			7,759	78.64%	215	2.18%
3 Arizona	85,470	81,956	95.89%			57,352	67.10%		
4 Arkansas	36,840	36,702	99.63%					599	1.63%
5 California	532,610	527,297	99.00%					3,392	1%
6 Colorado	65,679	63,219	96.25%	60,530	92.16%				
7 Connecticut	43,370	43,722	* 100.81%					219	0.50%
8 Delaware	11,639	11,714	* 100.64%	10,692	91.86%				
9 District of Columbia	15,159	15,125	99.78%					287	1.89%
10 Florida	204,305	204,030	99.87%						
11 Georgia	133,524	189,498	* 141.92%						
12 Hawaii	17,638	17,612	99.85%					197	1.12%
13 Idaho	19,863	20,254	* 101.97%			13,804	69.50%	199	1.00%
14 Illinois	181,984	188,164	* 103.40%						
15 Indiana	87,891	87,639	99.71%					31,782	36.16%
16 Iowa	38,418	38,141	99.28%					1,439	3.75%
17 Kansas	39,232	39,031	99.49%						
18 Kentucky	54,423	54,515	* 100.17%						
19 Maine	13,462	13,341	99.10%						
20 Maryland	69,574	72,390	* 104.05%	61,152	87.89%				
21 Massachusetts	82,673	82,703	* 100.04%					8,818	10.67%
22 Michigan	134,889	134,022	99.36%						
23 Minnesota	67,546	68,402	* 101.27%					319	0.47%
24 Mississippi	42,980	44,075	* 102.55%						
25 Missouri	78,302	78,640	* 100.43%					5,282	6.75%
26 Montana	10,927	10,825	99.07%			3,540	32.40%		
27 Nebraska	24,961	24,863	99.61%						
28 Nevada	30,387	30,659	* 100.90%	25,668	84.47%				
29 New Hampshire	13,987	13,879	99.23%			815	5.83%		
30 New Jersey	112,311	112,241	99.94%					10,716	9.54%
31 New Mexico	26,809	25,745	96.03%	953	3.55%			84	0.31%
32 New York	259,995	258,449	99.41%						
33 North Carolina	121,347	120,750	99.51%						
34 North Dakota	8,847	8,806	99.54%					494	5.58%
35 Ohio	155,943	160,566	* 102.96%					37,021	23.74%
36 Oklahoma	48,650	52,760	* 108.45%						
37 Oregon	46,790	46,879	* 100.19%	43,423	92.80%			862	1.84%
38 Pennsylvania	146,857	36,326	24.74%						
39 Rhode Island	13,180	13,150	99.77%					427	3.24%
40 South Carolina	53,562	52,176	97.41%					3,657	6.83%
41 South Dakota	10,589	10,220	96.52%			139	1.31%	42	0.40%
42 Tennessee	84,832	79,539	93.76%						
43 Texas	368,019	355,100	96.49%	348,132	94.60%			1,919	0.52%
44 Utah	48,454	47,423	97.87%	18,883	38.97%			406	0.84%
45 Vermont	6,277	6,040	96.22%					326	5.19%
46 Virginia	96,755	99,410	* 102.74%					12,594	13.02%
47 West Virginia	21,620	21,321	98.62%						
48 Wisconsin	68,250	66,614	97.60%			1,196	1.75%	1,230	1.80%
49 Wyoming	5,847	5,480	93.72%						
50 Puerto Rico	59,461	57,048	95.94%					22	0.04%
51 Virgin Islands	1,851	1,851	100.00%					13	0.70%
52 NeoGen Screening	144,319	140,789	97.55%			1,984	1.37%	1,546	1.07%
TOTAL	a 4,120,726	4,042,816	b	569,433	b	99,884	b	126,053	b

* Percentage > 100% denotes inability of program to separate newborns screened from specimens received; a = totals should be viewed as a rough estimate due to inability of many states, to know actual number of newborns screened. Many programs count sample received but cannot eliminate duplication when multiple samples are received; b = cannot be calculated due to inaccurate data.

6. Maple Syrup Urine Disease (MSUD)

6.0 Introduction

Screening for maple syrup urine disease (MSUD) began in some screening programs in the 1960's although most programs now screening developed their programs later. The relatively low incidence of MSUD has made it a controversial disorder for inclusion in many newborn screening programs. Those that screen uniformly use the bacterial assay for leucine and even though there are relatively few programs screening, there is some disagreement on screening cutoff levels and definitions. All data reported here was collected from questionnaires submitted to programs as noted in Chapter 3. Only programs noting MSUD as included in their testing scheme are listed in the Tables.

6.1 Definitions for Maple Syrup Urine Disease

Programs were asked to give the definition used by their program to define maple syrup urine disease. There is still not general agreement among programs as to the level of leucine to be used for defining possible cases of MSUD, either 2 mg/dL or 4 mg/dL. It is hoped that eventually programs can come to a consensus as to the best definition to use and the extent to which a metabolic specialist should be involved in the final diagnosis. From the definitions submitted, it is the usual case that a specialist is consulted to make the final diagnosis and program guidance is limited.

6.2 Laboratory Techniques

In order to identify laboratory techniques in use, programs were asked to report their laboratory protocols. These data are reported in Table 6.02.

6.3 Initial Screening Results

In order to ascertain the effectiveness of screening for MSUD, programs were asked to report their findings on initial screening. Initial

screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of infants screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated.

Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 6.03. Programs wishing to explain some of their responses included information as footnotes to the table. Because programs continue to report inaccurate or inappropriate data in some of their responses, no totals were given in some columns of the table. Care should be taken in using these data, since not all programs reported data.

6.4 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when a second test is performed without regard for the initial screening result. Programs that requested a second screen because of either a certain result on the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as 'repeat screens.'

For each case, the data requested included the total infants tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. It should be recognized that this is a different way of reporting these data than previously and may, therefore, be somewhat confusing. It is hoped that these data better represent the findings from additional screens. Once again, the reader is cautioned about using the data without regard to notes of explanation.

6.5 Cases of MSUD Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of MSUD, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 6.05. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

6.6 Time Until Treatment

In order to look at overall program efficiency, programs were asked to report the number of days from birth until treatment of MSUD. The definition of treatment used in the questionnaire was “initiation of dietary or drug regimen” and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 6.06.

6.7 Historic Data

In order to document the value of screening for MSUD, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of cases detected over the years. These data are given in Table 6.07.

6.8 Total Newborns Screened

Table 6.08 gives a summation of the data contained in Tables 6.03 and 6.04 along with the number of births in each state or territory as reported in Table 1.01. This table was included so that tabulation of these data could be viewed without having to refer to several tables.

6.9 Summation of Results

Table 6.09 sums the significant testing data from this Chapter. It includes the total births from Table 1.01, the total infants reported as being screened, and the testing data reported in Tables 6.03 and 6.04. Comparative percentages have been provided at the request of a number of readers, but in many cases these data are based on estimates or on data that appear to contain duplicate information, thus total compliance percentages may exceed 100% in some cases.

Table 6.01: Definitions for Maple Syrup Urine Disease

State/Territory	Definition for Maple Syrup Urine Disease
1 Alaska	Branched chain Amino Acids elevated above normal on diet and infant symptomatic.
2 Arizona	Serum leucine \geq 4 mg/dL.
3 Connecticut	Classified by treatment center.
4 District of Columbia	Elevated leucine.
5 Georgia	Branched chain amino acids significantly elevated confirmed by plasma amino acid analysis.
6 Hawaii	Branched chain amino acids elevated above normal and infant symptomatic.
7 Idaho	Branched chain Amino Acids elevated above normal on diet and infant symptomatic.
8 Indiana	Leucine \geq 4.0 mg/dL on screen. Confirmation by medical consultant and diagnostic tests.
9 Maine	Leucine < 6 mg/dL on screen with confirmation by medical consultant and/or reference lab performing diagnostic tests.
10 Maryland	Leucine >2 mg/dL on screen, elevated leucine, valine and isoleucine on amino acid profile.
11 Massachusetts	Diagnosis and classification determined by metabolic consultant.
12 Michigan	Deficiency in the enzyme BCKAD resulting in elevations in serum (leucine, isoleucine, valine).
13 Nevada	Branched chain amino acid elevated above normal on diet and symptomatic.
14 New Hampshire	Diagnosis and classification made by metabolic consultant.
15 New York	Diagnosis and classification determined by treatment center.
16 North Carolina	Diagnosis and classification made by medical consultant.
17 North Dakota	Diagnosis determined by medical consultant.
18 Oregon	Branched chain Amino Acids elevated above normal on diet and infant symptomatic.
19 Pennsylvania	An autosomal recessive disorder of branched chain ketoacid decarboxylation resulting in high body fluid levels of leucine.
20 Rhode Island	Diagnosis and classification determined by metabolic consultant.
21 Vermont	Persistent leucine elevation > 6 mg/dL.
22 Virginia	Branched-chain Keto acid decarboxylase deficiency resulting in elevated leucine levels.
23 Wyoming	Leucine > 400 increased allisoleucine, decreased branched chain ketoacid dehydrogenase in fibroblasts.
24 Virgin Islands	> 4 mg/dL.

Table 6.02: Maple Syrup Urine Disease - Laboratory Testing

State/Territory	Leucine BIA Assay	Other	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Immediate Serum Follow-up</i>
1 Alaska	X			≥4 mg/dL Leucine (all elevations require phone calls).
2 Arizona	X		None	≥ 4 mg/dL
3 Connecticut	X			≥ 4 mg/dL
4 District of Columbia		MS/MS		Leucine - > 400 μM/L.
5 Georgia	X		Leu ≥ 4 mg/dL without symptoms but < 6 mg/dL	Leucine ≥ 6 mg/dL or Leucine ≥4 mg/dL with symptoms.
6 Hawaii	X		> 4 mg/dL (all elev. Require phone calls) if on hyperal repeat filter paper	> 4 mg/dL if not on hyperal and symptomatic
7 Idaho	X			≥ 4 mg/dL % all elevations require phone notification.
8 Indiana	X		Leucine ≥ 4.0 < 5.9 mg/dL	Leucine ≥ 6.0 mg/dL.
9 Maine		MS/MS		
10 Maryland	X	HPLC	> 2 mg/dL and < 4 mg/dL	> 4 mg/dL
11 Massachusetts		MS/MS	≥ 6 mg/dL	≥ 6 mg/dL, all elevations reported by phone.
12 Michigan		a	5.5 - 6.9 if ≤ 8 days old	≥ 7.0 if < 8 days old, > 8.0 if > 8 days old.
13 Nevada	X		Leucine 4 mg/dL	Leucine ≥ 4 mg/dL
14 New Hampshire		MS/MS	≥ 6 mg/dL	≥ 6 mg/dL, All elevations reported by phone.
15 New York	X		≥4 mg/dL	> 4 mg/dL
16 North Carolina		MS/MS	≥ 3.51 mg/dL (300μM)	5.86 mg/dL (500 μM)
17 North Dakota	X		≥ 4 mg/dL	≥ 6 - 8 mg/dL
18 Oregon	X			≥4 mg/dL (all elevations require phone calls).
19 Pennsylvania		MS/MS		
20 Rhode Island	X		≥ 5 mg/dL	> 6 mg/dL
21 Vermont		MS/MS	< 6 mg/dL	> 6 mg/dL
22 Virginia	X	HPLC	≥ 4 mg/dL	≥ 6 mg/dL
23 Wyoming	X		DA	Leucine ≥ 4 mg/dL
24 Virgin Islands	X		> 4 mg/dL	

a = fluorometric since 6-14-00.

Table 6.03: Initial Screening Results - Maple Syrup Urine Disease

State/Territory	Number of Newborns Screened	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number of Newborns Confirmed with MSUD
1 Alaska	9,821	0		0
2 Arizona	81,956	4	0	1
3 Connecticut	43,722	32	0	0
4 District of Columbia	15,125	0	NA	0
5 Georgia	189,498	0	0	2
6 Hawaii	17,612	5	2 c	0
7 Idaho	20,254	2	0	0
8 Indiana	87,639	68	3	1
9 Maine	13,341	0	0	0
10 Maryland	72,390	108	4	2
11 Massachusetts	82,703	37	0	0
12 Michigan	134,022	120	0	0
13 Nevada	30,659	5	0	0
14 New Hampshire	13,879	4	0	0
15 New York	258,449	131	ND	0
16 North Carolina	114,389	NC	NC	0
17 North Dakota	9,815	1	0	0
18 Oregon	46,879	5	0	0
19 Pennsylvania	145,874	6	0	2
20 Rhode Island	13,150	0	0	0
21 Vermont	6,040	0	0	0
22 Virginia	99,410	42	0	0
23 Wyoming	~ 1,530	0	0	0
24 Virgin Islands	1,851	8	0	0
TOTAL	a	a	a	8 b

a = totals not given - too many programs reported inaccurate or inappropriate data; **b** = total given does not include all states; **c** = expired.

Table 6.04: Second Screens for Maple Syrup Urine Disease

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed with MSUD			
	R	D	P		R	D	P	Total	R	D	P	Total	R	D	P	Total
1 Alaska		7,759	216	7,975		0		0								
2 Arizona		57,352		57,352		3		3								
3 Connecticut			235	235			2	2		0		0		0		0
4 District of Columbia			287	287				0	0		0					0
5 Georgia			ND	ND												
6 Hawaii			189	189												
7 Idaho		13,804	199	14,003												
8 Indiana			DA	31,782			25	25			2	2				0
9 Maine																
10 Maryland	61,152			61,152	7			7	5			5				
11 Massachusetts			8,818	8,818			50	50				0				0
12 Michigan				0			0	0			0	0		1		1
13 Nevada	25,668			25,668												
14 New Hampshire		815		815												
15 New York			DA	0		DA	N/C			DA	N/C			DA	N/C	
16 North Carolina																
17 North Dakota			DA	2,647		DA	1	1		DA	0	0		DA	0	0
18 Oregon	43,423		864	44,287	1		0	1								
19 Pennsylvania																
20 Rhode Island		N/C	N/C	427												
21 Vermont				326												
22 Virginia				12,648			12	12		0	0	0		0	0	0
23 Wyoming			DA	NC		DA	N/C			DA	N/C					
24 Virgin Islands				8												
Total	a	130,243	79,730	58,646	b			b				b	0	1	0	1 c

a = totals given reflect program responses and should be viewed as estimates only, given the number not reporting and the number of caveats listed;

b = totals not given - too many programs reported inaccurate or inappropriate data; **c** = totals given do not include all states.

Table 6.05: Cases of Maple Syrup Urine Disease

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic				
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total	
1 Alaska																			
2 Arizona									1				1	0	1				
3 Connecticut																			
4 District of Columbia																			
5 Georgia		1											0	1	2	³	1	1	
6 Hawaii																			
7 Idaho																			
8 Indiana		1											0	1	1				
9 Maine																			
10 Maryland	1			1									1	1	2				
11 Massachusetts																			
12 Michigan	1												1	0	1				
13 Nevada																			
14 New Hampshire																			
15 New York																			
16 North Carolina																			
17 North Dakota																			
18 Oregon																			
19 Pennsylvania	1	1											1	1	2				
20 Rhode Island																			
21 Vermont																			
22 Virginia																			
23 Wyoming																			
24 Virgin Islands																			
TOTAL	3	3	0	1	0	0	0	0	1	0	0	0	4	4	9	0	1	1	

¹did not report a breakdown of Race/Ethnicity.

²total reported is from Table 6.03 and Table 6.04; therefore total does not equal or may differ from data given in Table 6.05 and denotes a problem in reliability.

³Ethnicity counted as a Race and is included in total

Table 6.06: Days from Birth Until Treatment Initiated for Maple Syrup Urine Disease
Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	TOTAL
1 Alaska																0
2 Arizona					1											1
3 Connecticut																0
4 District of Columbia																0
5 Georgia													2			2
6 Hawaii																0
7 Idaho																0
8 Indiana							1									1
9 Maine																0
10 Maryland					1	1										2
11 Massachusetts																0
12 Michigan														1		1
13 Nevada																0
14 New Hampshire																0
15 New York																0
16 North Carolina																0
17 North Dakota																0
18 Oregon																0
19 Pennsylvania	1						1									2
20 Rhode Island																0
21 Vermont																0
22 Virginia																0
23 Wyoming																0
24 Virgin Islands																0

Table 6.07: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Maple Syrup Urine Disease		
	Year Started	Total Years	Total Cases
1 Alaska	1987	13	0
2 Arizona	1994	6	2
3 Connecticut	1993	7	0
4 District of Columbia	1996	4	0
5 Georgia	Sep-78	21 1/4	19
6 Hawaii	Jul-97	2 1/2	2
7 Idaho	1962	38	0
8 Indiana	Jul-85	15 1/2	2
9 Maine	1976	24	0
10 Maryland	1972	28	14
11 Massachusetts	1963	37	19
12 Michigan	1987	13	8
13 Nevada NR			
14 New Hampshire	1983	17	0
15 New York	1968	32	26
16 North Carolina NR			
17 North Dakota	1992	8	0
18 Oregon NR			
19 Pennsylvania NR			
20 Rhode Island NR			
21 Vermont	Jul-89	11 1/2	0
22 Virginia	1984	16	3
23 Wyoming	Apr-79	17 3/4	0
24 Virgin Islands NR			

Table 6.08: Total Newborns Screened for Maple Syrup Urine Disease

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number of Newborns Confirmed with MSUD
1 Alaska	9,866	0	0	0
2 Arizona	85,470	7	0	1
3 Connecticut	43,370	34	0	0
4 District of Columbia	15,159	0	0 N/A	0
5 Georgia	133,524	0	0	2
6 Hawaii	17,638	5	2 b	0
7 Idaho	19,863	2	0	0
8 Indiana	87,891	93	5	1
9 Maine	13,462	0	0	0
10 Maryland	69,574	115	9 c	2
11 Massachusetts	82,673	87	0	0
12 Michigan	134,889	120	0	1
13 Nevada	30,387	5	0	0
14 New Hampshire	13,987	4	0	0
15 New York	259,995	131	N/D	0
16 North Carolina	121,347	0	0	0
17 North Dakota	8,847	2	0	0
18 Oregon	46,790	6	0	0
19 Pennsylvania	146,857	6	0	2
20 Rhode Island	13,180	0	0	0
21 Vermont	6,277	0	0	0
22 Virginia	96,755	54	0	0
23 Wyoming	5,847	0	0	0
24 Virgin Islands	1,851	8	0	0
TOTAL	1,465,499	679	16	9 a

a = totals given do not include all states; **b** = expired; **c** = 4 expired.

Table 6.09: Summation Results of Testing for Maple Syrup Urine Disease

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screening calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary 2nd Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Alaska	9,866	9,821	99.54%			7,759	78.64%	216	2.19%
2 Arizona	85,470	81,956	95.89%			57,352	67.10%		
3 Connecticut	43,370	43,722	* 100.81%					235	0.54%
4 District of Columbia	15,159	15,125	99.78%					287	1.89%
5 Georgia	133,524	189,498	* 141.92%						
6 Hawaii	17,638	17,612	99.85%					189	1.07%
7 Idaho	19,863	20,254	* 101.97%			13,804	69.50%	199	1.00%
8 Indiana	87,891	87,639	99.71%					31,782	36.16%
9 Maine	13,462	13,341	99.10%						
10 Maryland	69,574	72,390	* 104.05%	61,152	87.89%				
11 Massachusetts	82,673	82,703	* 100.04%					8,818	10.67%
12 Michigan	134,889	134,022	99.36%						
13 Nevada	30,387	30,659	* 100.90%	25,668	84.47%				
14 New Hampshire	13,987	13,879	99.23%			815	5.83%		
15 New York	259,995	258,449	99.41%						
16 North Carolina	121,347	114,389	94.27%						
17 North Dakota	8,847	9,815	* 110.94%					2,647	29.92%
18 Oregon	46,790	46,879	* 100.19%	43,423	92.80%			864	1.85%
19 Pennsylvania	146,857	145,874	99.33%						
20 Rhode Island	13,180	13,150	99.77%					427	3.24%
21 Vermont	6,277	6,040	96.22%					326	5.19%
22 Virginia	96,755	99,410	* 102.74%					12,648	13.07%
23 Wyoming	5,847	1,530	26.17%						
24 Virgin Islands	1,851	1,851	100.00%					8	0.43%
TOTAL	a 1,465,499	1,510,008	b	130,243	b	79,730	b	58,646	b

* Percentage > 100% denotes inability of program to separate newborns screened from specimens received; **a** = totals should be viewed as a rough estimate due to inability of many states, to know actual number of newborns screened. Many programs count samples received but cannot eliminate duplication when multiple samples are received; **b** = cannot calculate due to inaccurate data.

7. Homocystinuria

7.0 Introduction

Screening for homocystinuria began in Massachusetts in 1968, but most programs now screening developed their programs in the 1970's or later. The relatively low incidence of homocystinuria and questions about the laboratory test used and when the test is most accurate have made homocystinuria a controversial disorder for inclusion in many newborn screening programs. Those that screen uniformly use the bacterial assay for methionine and even though there are relatively few programs screening, there is some disagreement on screening cutoff levels and definitions. All data reported here was collected from questionnaires submitted to programs as noted in Chapter 3. Only programs with homocystinuria screening included in their testing scheme are listed in the Tables.

7.1 Definitions for Homocystinuria

Programs were asked to give the definition used by their program to define possible cases of homocystinuria. In this case, there is general agreement among programs that a methionine level in excess of 2 mg/dL defines a presumptive positive screen and that a referral should be made to a metabolic specialist. The results of the questionnaire responses are given in Table 7.01.

7.2 Laboratory Procedures

In order to identify laboratory procedures in use, programs were asked to report their laboratory protocols and definitions for samples considered 'not normal.' These data are reported in Table 7.02.

7.3 Initial Screening Results

In order to ascertain the effectiveness of screening for homocystinuria, programs were asked to report their findings on initial screening.

Initial screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of newborns screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated. Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 7.03. Programs wishing to explain some of their responses included information as footnotes to the table. Because programs continue to report inaccurate or inappropriate data in some of their responses, no totals were given in some columns of the table. Some care should be taken in using these data, since not all programs reported data.

7.4 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are reported in Table 7.04 and are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when a second test is performed without regard for the initial screening result. Programs that requested a second screen because of either a certain result on the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as 'repeat screens' and these are also included.

For each case, the data requested included the total newborns tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. It should be recognized that this is a different way of reporting these data than previously and may, therefore, be somewhat confusing. It is hoped that these data better represent the findings from additional screens. Once again, the reader is cautioned about using the data without regard to notes of explanation.

7.5 Cases of Homocystinuria Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of homocystinuria, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 7.05. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

7.6 Time Until Treatment

In order to look at overall program efficiency, programs were asked to report the number of days from birth until treatment of homocystinuria. The definition of treatment used in the questionnaire was “initiation of dietary or drug regimen” and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 7.06.

7.7 Historic Data

In order to document the value of screening for homocystinuria, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of cases detected over the years. These data are given in Table 7.07.

7.8 Total Newborns Screened

Table 7.08 gives a summation of the data contained in Tables 7.03 and 7.04 along with the number of births in each state or territory as reported in Table 1.01. This table was included so that a tabulation of these data could be viewed without having to refer to several tables.

7.9 Summation of Results

Table 7.09 sums the significant testing data from this Chapter. It includes the total births from Table 1.01, the total newborns reported as being screened, and the testing data reported in Tables 7.03 and 7.04. Comparative percentages have been provided at the request of a number of readers, but in many cases these data are based on estimates or on data that appear to contain duplicate information, thus total compliance percentages may exceed 100% in some cases. Data responders are requested to review these data and adjust their responses to future questionnaires so that more meaning can be associated with these data. In particular, the data in this table seem to point out the difficulty of many programs in separating total samples received from total births screened (i.e. many cannot differentiate between first tests received and others that might have been submitted as independent second tests or repeat screens).

Table 7.01: Definitions for Homocystinuria

State/Territory	Definition for Homocystinuria
1 Arizona	Serum methionine level > 2 mg/dL.
2 Connecticut	Classified by treatment center.
3 District of Columbia	Elevated methionine.
4 Georgia	Presence of homocystinuria & significantly elevated methionine in plasma amino acid analysis.
5 Indiana	Methionine \geq 3.0 mg/dL on screen. Confirmation by medical consultant and diagnostic tests.
6 Maine	Persistent methionine levels > 1.5 mg/dL confirmed by medical consultant and/or reference lab performing diagnostic tests.
7 Maryland	Elevated methionine > 2 mg/dL.
8 Massachusetts	Diagnosis and classification determined by metabolic consultant.
9 New Hampshire	Diagnosis and classification made by metabolic consultant.
10 New York	Diagnosis and classification made by treatment center.
11 North Carolina	Diagnosis and classification made by medical consultant.
12 Ohio	Diagnosis and classification made by medical consultant.
13 Rhode Island	Diagnosis and classification determined by metabolic consultant.
14 Vermont	Persistent methionine elevation > 1.5 mg/dL
15 Virginia	Cystathionine B-synthetase deficiency resulting in elevated methionine levels.
16 Wyoming	Elevated methionine decreased cystathionine beta synthase in fibroblasts. Elevated homocystine in older children.
17 Virgin Islands	> 2 mg/dL.
18 NeoGen Screening	Elevated methionine.

Table 7.02: Homocystinuria - Laboratory Testing

State/Territory	Methionine BIA Assay	Other	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Immediate Serum Follow-up</i>
1 Arizona	X		1 abnormal screen between 2 and 4	2 abnormal screens or 1 abnormal > 4 mg/dL
2 Connecticut	X		Methionine 3 - 6 mg/dL	Methionine > 6 mg/dL
3 District of Columbia		MS/MS		Methionine > 60 µM/L
4 Georgia	X		Methionine ≥ 2 mg/dL but < 4 mg/dL	Methionine ≥ 4 mg/dL
5 Indiana	X		Methionine 2 - 2.9 mg/dL	Methionine ≥ 3 mg/dL
6 Maine		MS/MS	Methionine > 1.5 mg/dL	
7 Maryland	X	HPLC	Methionine > 1 mg/dL and < 2 mg/dL	Methionine > 2 mg/dL
8 Massachusetts		MS/MS	Methionine ≥ 1.5 mg/dL	Methionine ≥ 1.5 mg/dL, all elevations are phoned.
9 New Hampshire		MS/MS	Methionine ≥ 1.5 mg/dL	
10 New York	X		Methionine ≥ 1.5 mg/dL and < 4 mg/dL	Methionine ≥ 4 mg/dL
11 North Carolina		MS/MS	Methionine ≥ 1.57 mg/dL (105 µM)	Methionine ≥ 4.48 mg/dL (300µM)
12 Ohio	X		Methionine ≥ 2 mg/dL	Methionine ≥ 2 mg/dL
13 Rhode Island	X		Methionine ≥ 1 mg/dL	Methionine ≥ 1.5 mg/dL
14 Vermont		MS/MS	Methionine > 1 - 1.5 mg/dL	Methionine > 1.5 mg/dL
15 Virginia	X		Methionine ≥ 2 mg/dL - 3 mg/dL	Methionine ≥ 4 mg/dL
16 Wyoming	X	HPLC	Methionine ≥ 2 mg/dL	Methionine ≥ 4 mg/dL
17 Virgin Islands	X		Methionine > 2 mg/dL	
18 NeoGen Screening		MS/MS		

Table 7.03: Initial Screening Results - Homocystinuria

State/Territory	Number of Newborns screened	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number of Newborns Confirmed with Homocystinuria
1 Arizona	81,956	19	0	0
2 Connecticut	43,722	8	0	0
3 District of Columbia	15,125	3	0	0
4 Georgia	189,498	0	0	0
5 Indiana	87,639	99	4	0
6 Maine	13,341	4	0	0
7 Maryland	72,352	38	0	0
8 Massachusetts	82,703	42	N/A	0
9 New Hampshire	13,879	7	0	0
10 New York	258,449	241	N/D	3
11 North Carolina	120,750	NC	NC	0
12 Ohio	160,566	289	0	1
13 Rhode Island	13,150	0	0	0
14 Vermont	6,040	2	0	0
15 Virginia	99,410	10	0	0
16 Wyoming	~ 1,530	2	0	0
17 Virgin Islands	1,851	5	0	0
18 NeoGen Screening	140,789	24	3 a	1
TOTAL				5 b

a = physician notification; **b** = total given does not include all states.

Table 7.04: Second Screens for Homocystinuria

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed with Homocystinuria			
	R	D	P		R	D	P	Total	R	D	P	Total	R	D	P	Total
1 Arizona		57,352		57,352	24			24	0	0	0		0	0	0	
2 Connecticut		41,601		41,601	5			5	0		0		0		0	
3 District of Columbia			287	287			1	1			1	a			0	
4 Georgia			ND	ND												
5 Indiana			DA	31,782			12	12			2	2			0	
6 Maine															0	
7 Maryland	61,152			61,152	5			5	2		2					
8 Massachusetts			8,818	8,818			50	50			0				0	
9 New Hampshire		815		815												
10 New York			DA	0		DA	N/C			DA	N/C		DA	N/C		
11 North Carolina																
12 Ohio			37,021	37,021				0						0	0	
13 Rhode Island			427	427												
14 Vermont			326	326			0	0								
15 Virginia			12,645	12,645			5	5		0	0	0		0	0	
16 Wyoming	DA	DA	N/C		DA	DA	N/C		DA	DA	N/C		DA	DA	N/C	
17 Virgin Islands			5	5				0			0				0	
18 NeoGen Screening		1,984	1,546	3,530				0			0				0	
Total	b	61,152	101,752	92,857	d			d			d	0	0	0	0	

a = lab lost to follow-up - DC MCH has more info; **b** = totals given reflect program responses should be viewed as estimates only, given the number not reporting and the number of caveats listed; **c** = these numbers are overall; **d** = totals not given - too many programs reported inaccurate or inappropriate or inappropriate data; **e** = totals given does not include all states; **f** = 5 confirmed with hypermethioninemia.

Table 7.05: Cases of Homocystinuria

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic				
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total	
1 Arizona																			
2 Connecticut																			
3 District of Columbia																			
4 Georgia																			
5 Indiana																			
6 Maine																			
7 Maryland																			
8 Massachusetts																			
9 New Hampshire																			
10 New York			2							1				2	1	3			
11 North Carolina																			
12 Ohio												1	0	1	1				
13 Rhode Island																			
14 Vermont																			
15 Virginia																			
16 Wyoming																			
17 Virgin Islands																			
18 NeoGen Screening	1												1	0	1				
TOTAL	1	0	2	0	0	0	0	0	0	1	0	1	3	2	5				

¹did not report a breakdown of Race/Ethnicity.

Table 7.06: Days from Birth Until Treatment Initiated for Homocystinuria
 Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	TOTAL
1 Arizona																
2 Connecticut																
3 District of Columbia																
4 Georgia																
5 Indiana																
6 Maine																
7 Maryland																
8 Massachusetts																
9 New Hampshire																
10 New York													1	2		3
11 North Carolina																
12 Ohio															1	1
13 Rhode Island																
14 Vermont																
15 Virginia																
16 Wyoming																
17 Virgin Islands																
18 NeoGen Screening															1	1

Table 7.07: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Homocystinuria		
	Year Started	Total Years	Total Cases
1 Arizona	1994	6	2
2 Connecticut	1993	7	0
3 District of Columbia	1996	4	0
4 Georgia	Sep-78	21 1/4	6
5 Indiana	Jul-85	15 1/2	1
6 Maine	1976	24	3
7 Maryland	1972	28	8
8 Massachusetts	1968	32	8
9 New Hampshire	1983	17	4
10 New York	1975	25	18
11 North Carolina NR			
12 Ohio NR			
13 Rhode Island NR			
14 Vermont	Jul-89	11 1/2	0
15 Virginia NR			
16 Wyoming	Apr-79	17 3/4	0
17 Virgin Islands NR			
18 NeoGen Screening NR			

Table 7.08: Total Newborns Screened for Homocystinuria

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number of Newborns Confirmed with Homocystinuria
1 Arizona	85,470	43	0	0
2 Connecticut	43,370	13	0	0
3 District of Columbia	15,159	4	1	0
4 Georgia	133,524	0	0	0
5 Indiana	87,891	111	6	0
6 Maine	13,462	4	0	0
7 Maryland	69,574	43	2	0
8 Massachusetts	82,673	92	0	0
9 New Hampshire	13,987	7	0	0
10 New York	259,995	241	N/D	3
11 North Carolina	121,347	0	0	0
12 Ohio	155,943	289	0	1
13 Rhode Island	13,180	0	0	0
14 Vermont	6,277	2	0	0
15 Virginia	96,755	15	0	0
16 Wyoming	5,847	2	0	0
17 Virgin Islands	1,851	5	0	0
18 NeoGen Screening	144,319	24	3	1
TOTAL	1,350,624	895	12	5 a

a = total given does not include all state.

Table 7.09: Summation Results of Testing for Homocystinuria

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screen calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Arizona	85,470	81,956	95.89%			57,352	67.10%		
2 Connecticut	43,370	43,722	* 100.81%			41,601	95.92%		
3 District of Columbia	15,159	15,125	99.78%					287	1.89%
4 Georgia	133,524	189,498	* 141.92%						
5 Indiana	87,891	87,639	99.71%					31,782	36.16%
6 Maine	13,462	13,341	99.10%						
7 Maryland	69,574	72,352	* 103.99%	61,152	87.89%				
8 Massachusetts	82,673	82,703	* 100.04%					8,818	10.67%
9 New Hampshire	13,987	13,879	99.23%			815	5.83%		
10 New York	259,995	258,449	99.41%						
11 North Carolina	121,347	120,750	99.51%						
12 Ohio	155,943	160,566	* 102.96%					37,021	23.74%
13 Rhode Island	13,180	13,150	99.77%					427	3.24%
14 Vermont	6,277	6,040	96.22%					326	5.19%
15 Virginia	96,755	99,410	* 102.74%					12,645	13.07%
16 Wyoming	5,847	1,530	26.17%						
17 Virgin Islands	1,851	1,851	100.00%					5	0.27%
18 NeoGen Screening	144,319	140,789	97.55%			1,984	1.37%	1,546	1.07%
TOTAL	a	1,350,624	b	61,152	b	101,752	b	92,857	b

* Percentage > 100% denotes inability of program to separate newborns screened from specimens received; **a** = totals should be viewed as a rough estimate due to inability of many states to know actual number of newborns screened. Many programs count samples received but cannot eliminate duplication when multiple samples are received; **b** = cannot be calculated due to inaccurate data.

8. Biotinidase Deficiency

8.0 Introduction

Screening for biotinidase deficiency is relatively young, having started only in the mid-1980's in most programs. Screening involves the use of a semi-quantitative measurement of biotinidase activity and treatment is usually doses of oral biotin. Most screening programs use the same semi-quantitative assay although variations may exist in the degrees of automation. There is little disagreement on screening cutoff levels and definitions. All data reported here was collected from questionnaires submitted to programs as noted in Chapter 3. Only programs with biotinidase deficiency screening included in their testing scheme are listed in the Tables.

8.1 Definitions for Biotinidase Deficiency

Programs were asked to give the definition used by their program to define possible cases of biotinidase deficiency. In this case, there is general agreement among programs that decreased enzyme activity signals the presence of biotinidase deficiency, although there is some disagreement as to the exact level that is 'not normal.' It is generally agreed that an abnormal screening result should result in referral to a metabolic specialist. The results of the questionnaire responses are given in Table 8.01.

8.2 Laboratory Procedures

In order to identify laboratory procedures in use, programs were asked to report laboratory protocols and definitions for samples considered 'not normal.' These data are reported in Table 8.02.

8.3 Initial Screening Results

In order to ascertain the effectiveness of screening for biotinidase deficiency, programs were asked to report their findings on initial screening. Initial screening was defined as the

first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of newborns screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated.

Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 8.03. Programs wishing to explain some of their responses included information as footnotes to the table. Because programs continue to report inaccurate or inappropriate data in some of their responses, no totals were given in some columns of the table. Some care should be taken in using these data, since not all programs reported.

8.4 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are reported in Table 8.04 and are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when a second test is performed without regard for the initial screening result. Programs that requested a second screen because of either a certain result on the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as 'repeat screens' and these are also included. For each case, the data requested included the total newborns tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. It should be recognized that this is a different way of reporting these data than previously and may, therefore, be somewhat confusing. It is hoped that these data better represent the findings from additional screens. Once again, the reader is cautioned about using the data without regard to notes of explanation.

8.5 Cases of Biotinidase Deficiency Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of biotinidase deficiency, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 8.05. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

8.6 Time Until Treatment

In order to look at overall program efficiency, programs were asked to report the number of days from birth until treatment of biotinidase deficiency. The definition of treatment used in the questionnaire was “initiation of dietary or drug regimen” and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 8.06.

8.7 Historic Data

In order to document the value of screening for biotinidase deficiency, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of cases detected over the years. These data are given in Table 8.07.

8.8 Total Newborns Screened

Table 8.08 gives a summation of the data contained in Tables 8.03 and 8.04 along with the number of births in each state or territory as reported in Table 1.01. This table was included so that a tabulation of these data could be viewed without having to refer to several tables.

8.9 Summation of Results

Table 8.09 sums the significant testing data from this Chapter. It includes the total births from Table 1.01, the total newborns reported as being screened, and the testing data reported in Tables 8.03 and 8.04. Comparative percentages have been provided at the request of a number of readers, but in many cases these data are based on estimates or on data that appear to contain duplicate information, thus total compliance percentages may exceed 100% in some cases.

Table 8.01: Definitions for Biotinidase Deficiency

State/Territory	Definition for Biotinidase Deficiency
1 Alaska	Enzyme activity partially or profoundly deficient.
2 Arizona	Zero enzyme activity.
3 Colorado	Confirmed low quantitative serum biotinidase activity.
4 Connecticut	Classified by treatment center.
5 Hawaii	Enzyme activity partially or profoundly deficient.
6 Idaho	Enzyme activity partially or profoundly deficient.
7 Illinois	Diagnosis and classification determined by pediatric medical consultant.
8 Indiana	The absence of color. Confirmed by quantitative level by medical consultant.
9 Louisiana	< 10% enzyme activity (profound), 10-30% enzyme activity (partial).
10 Maine	Enzyme activity partially or profoundly deficient. Confirmed by medical consultant.
11 Maryland	Decreased or absent biotinidase.
12 Massachusetts	Diagnosis and classification determined by metabolic consultant.
13 Michigan	Complete or partial biotinidase enzyme deficiency resulting in impaired biotin utilization.
14 Nebraska	Enzyme activity partially or profoundly deficient (wolfe).
15 Nevada	Enzyme activity partially or profound deficient.
16 New Mexico	Color change < 30% activity control.
17 New York	Diagnosis and classification determined by treatment center.
18 Oregon	Enzyme activity partially or profoundly deficient (Wolfe).
19 Rhode Island	Diagnosis and classification determined by metabolic consultant.
20 Vermont	Activity persistently \leq 10 % normal activity.
21 Virginia	Decreased or absent biotinidase enzyme.
22 Wisconsin	Diagnosis and classification made by treatment center.
23 Wyoming	Confirmed low quantitative serum biotinidase activity.
24 NeoGen Screening	Decreased or absent biotinidase activity (confirmed by quantitative biotinidase assay).

Table 8.02: Biotinidase Deficiency - Laboratory Testing

State/Territory	Testing Method	Other Method	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Immediate Serum Follow-up</i>
1 Alaska	Enzyme / Colorimetric		1st specimen has no color.	1st & 2nd specimens have no color.
2 Arizona	Colorimetric		Absent/reduced color on first.	Absence of color on 2nd screen.
3 Colorado	Wolf Colorimetric		<30% nml adult activity on initial specimen	<30% normal adult activity on initial and recall specimen.
4 Connecticut	Colorimetric			≥20%
5 Hawaii	Enzyme/Colorimetric		Activity absent on one specimen.	Activity absent on two specimens.
6 Idaho	Enzyme / Colorimetric		One specimen has no color.	2nd specimen together w/1st has no color.
7 Illinois	Colorimetric		Absence of color	Absence of color
8 Indiana	Wolf Qualitative		Decreased absence of color.	Abnormal filter paper x 2.
9 Louisiana	Colorimetric		Very light purple color.	No color development.
10 Maine	Colorimetric		< 30%	
11 Maryland	Colorimetric		No purple color on one testing.	Two abnormal filter paper testings on different samples.
12 Massachusetts	Enzyme / Colorimetric		< 30% enzyme activity.	< 20 % enzyme activity.
13 Michigan	Enzyme / Colorimetric		Very little color development.	Straw or no color.
14 Nebraska	Colorimetric-qualitative			visual comparison ≤ 30%
15 Nevada	Enzyme		On filter paper- 1 spec has no color.	2nd spec together with 1st has no color.
16 New Mexico	Enzyme		< 30% control.	Between 30% - 50% activity.
17 New York	Wolf Colorimetric		Very little color development.	No color development.
18 Oregon	Enzyme / Colorimetric		1st specimen has no color.	2nd specimens together w/1st has no color.
19 Rhode Island	Enzyme / Colorimetric		< 30% enzyme activity	< 20 % enzyme activity
20 Vermont	RIA		10% normal	10-30% activity.
21 Virginia	Colorimetric-qualitative		Decreased or the absence of biotinidase activity.	Two consecutive samples w/decreased or the absence of biotinidase activity.
22 Wisconsin	Colorimetric-qualitative		No color.	DA
23 Wyoming	Wolf Colorimetric		<30% normal adult activity on initial spec.	<30% normal adult activity/initial & recall.
24 NeoGen Screening	Enzyme/Colorimetric		Decreased or absence of biotinidase activity	Decreased or absence of biotinidase activity on second sample.

Table 8.03: Initial Screening Results - Biotinidase Deficiency				
State/Territory	Number Newborns Screened	Number Newborns with <u>NOT NORMAL</u> Test Results	Number Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number Newborns Confirmed with Biotinidase Deficiency
1 Alaska	9,821	8	0	0
2 Arizona	81,956	4	0	0
3 Colorado	63,219	14	0	0
4 Connecticut	43,727	1	0	1
5 Hawaii	17,612	9	9 a	0
6 Idaho	20,254	4	0	0
7 Illinois	188,164	16 e	0	1
8 Indiana	37,450	2	1	0
9 Louisiana	67,843	34	0	0
10 Maine	13,341	0	0	0
11 Maryland	72,390	18	0	0
12 Massachusetts	82,703	17	0 NA	0
13 Michigan	134,022	52	0	0
14 Nebraska	24,863	2	0	0
15 Nevada	30,659	7	0	0
16 New Mexico	25,745	39		1
17 New York	258,449	4	ND	2
18 Oregon	46,879	7	0	3
19 Rhode Island	13,150	0	0	0
20 Vermont	6,040	2	0	0
21 Virginia	99,410	34	0	0
22 Wisconsin	66,614	5	0	0
23 Wyoming	5,480	0	0	0
24 NeoGen Screening	140,789	4	1 b	0
TOTAL	d	d	d	8 c

a = expired; **b** = physician notification; **c** = total given does not include all states; **d** = totals not given - too many programs reported inaccurate or inappropriate data; **e** = included 12 unsatisfactory submissions.

Table 8.04: Second Screens for Biotinidase Deficiency

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen R	Newborns Receiving Discretionary Second Screens D	Newborns Receiving Repeat Second Screens P	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed with Biotinidase Deficiency					
					R	D	P	Total	R	D	P	Total	R	D	P	Total		
					1 Alaska		7,759	215	7,974		8		8					
2 Arizona		57,352		57,352	0	0	0	0		0	0	0		0	0	0		0
3 Colorado	60,530		N/C	60,530	N/C		N/C		0		0	0	0	0		0		0
4 Connecticut			191	191		0		0		0		0		0		0		0
5 Hawaii			186	186	0			0	N/A		N/A		0		0		0	0
6 Idaho		13,804	199 N/A	14,003		0		0		0		0		0		0		0
7 Illinois			N/C	0														
8 Indiana			13,190	13,190														
9 Louisiana																		
10 Maine																		
11 Maryland	61,152			61,152	8			8				0	5					5
12 Massachusetts			8,818	8,818				0				0						0
13 Michigan				0			0	0			0	0		5	0	5	e	
14 Nebraska		DA	DA															
15 Nevada	25,668			25,668														
16 New Mexico	23,581		168	23,749				0										
17 New York		DA		0		DA	N/C			DA	N/C			DA	N/C			
18 Oregon	43,423		862	44,285	3		2	5										
19 Rhode Island			427	427														
20 Vermont			326	326			0	0										
21 Virginia			12,667	12,667			6	6		0	0	0		0				0
22 Wisconsin		1,196	5	1,201		0	0	0		0	0	0		0	0	0		0
23 Wyoming	DA	DA	N/C			DA	DA	N/C		DA	DA	N/C		DA	DA	N/C		
24 NeoGen Screening		1,984	1,546	3,530 a														
Total	b	214,354	82,095	38,800	c			c				c	5	5	0	10	d	

a = these numbers are overall; **b** = totals given reflect program responses and should be viewed as estimates only, given the number not reporting and the number of caveats listed;

c = totals not given - too many programs reported inaccurate or inappropriate data; **d** = total given does not include all states; **e** = partial.

Table 8.05: Cases of Biotinidase Deficiency

Total detected on initial and second screen

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic				
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total	
1 Alaska																			
2 Arizona																			
3 Colorado																			
4 Connecticut									1				1	0	1				
5 Hawaii																			
6 Idaho																			
7 Illinois															1	³	1		1
8 Indiana																			
9 Louisiana																			
10 Maine																			
11 Maryland	2	1									1	1	3	2	5				
12 Massachusetts																			
13 Michigan															5	¹			
14 Nebraska																			
15 Nevada																			
16 New Mexico									1				1	0	1				
17 New York	2												2	0	2				
18 Oregon	2	1											2	1	3				
19 Rhode Island																			
20 Vermont																			
21 Virginia																			
22 Wisconsin																			
23 Wyoming																			
24 NeoGen Screening																			
TOTAL	6	2	0	0	0	0	0	0	2	0	1	1	9	3	18	²	1	0	1

¹did not report a breakdown of Race/Ethnicity.

²total reported is from Table 8.03 and Table 8.04; therefore total does not equal or may differ from data given in Table 8.05 and denotes a problem in reliability.

³Ethnicity counted as a Race and is included in total.

Table 8.06: Days from Birth Until Treatment Initiated for Biotinidase Deficiency

Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	TOTAL
1 Alaska																0
2 Arizona																0
3 Colorado																0
4 Connecticut										1						1
5 Hawaii																0
6 Idaho																0
7 Illinois														1		1
8 Indiana																0
9 Louisiana																0
10 Maine																0
11 Maryland														2	3	5
12 Massachusetts																0
13 Michigan															5	5
14 Nebraska																0
15 Nevada																0
16 New Mexico														1		1
17 New York							1				1					2
18 Oregon													1	2		3
19 Rhode Island																0
20 Vermont																0
21 Virginia																0
22 Wisconsin																0
23 Wyoming																0
24 NeoGen Screening																0

¹did not report a breakdown of Days from Birth.

²total reported is from Table 8.03 and Table 8.04; therefore total does not equal or may differ from data given in Table 8.06 and denotes a problem in reliability.

Table 8.07: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Biotinidase Deficiency		
	Year Started	Total Years	Total Cases
1 Alaska	1987	0	1
2 Arizona	1994	6	5
3 Colorado	Apr-89	10 3/4	12
4 Connecticut	1993	7	5
5 Hawaii	Jul-97	2 1/2	2
6 Idaho	1976	24	4
7 Illinois	1986	14	17
8 Indiana	Aug-00	1/4	0
9 Louisiana NR			
10 Maine	1999	1	0
11 Maryland	1985	15	17
12 Massachusetts	1991	9	30
13 Michigan	1987	13	53
14 Nebraska	1987	13	17
15 Nevada NR			
16 New Mexico	1981	19	1
17 New York	1987	13	43
18 Oregon NR			
19 Rhode Island NR			
20 Vermont	Jul-89	11 1/2	0
21 Virginia	1984	16	20
22 Wisconsin	1991	9	7
23 Wyoming	Apr-89	20 3/4	2
24 NeoGen Screening NR			

Table 8.08: Total Newborns Screened for Biotinidase Deficiency

State/Territory	Total Births	Number of Newborns with <u>NOTNORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Lost to follow-up	Number of Newborns Confirmed with Biotinidase Deficiency
1 Alaska	9,866	16	0	0
2 Arizona	85,470	4	0	0
3 Colorado	65,679	14	0	0
4 Connecticut	43,370	1	0	1
5 Hawaii	17,638	9	9	0
6 Idaho	19,863	4	0	0
7 Illinois	181,984	16	0	1
8 Indiana	87,891	2	1	0
9 Louisiana	68,275	34	0	0
10 Maine	13,462	0	0	0
11 Maryland	69,574	26	0	5
12 Massachusetts	82,673	17	0	0
13 Michigan	134,889	52	0	5 b
14 Nebraska	24,961	2	0	0
15 Nevada	30,387	7	0	0
16 New Mexico	26,809	39	0	1
17 New York	259,995	4	N/D	2
18 Oregon	46,790	12	0	3
19 Rhode Island	13,180	0	0	0
20 Vermont	6,277	2	0	0
21 Virginia	96,755	40	0	0
22 Wisconsin	68,250	5	0	0
23 Wyoming	5,847	0	0	0
24 NeoGen Screening	144,319	4	1	0
TOTAL	1,604,204	310	11	18 a

a = total given does not include all states; **b** = 5 partial.

Table 8.09: Summation Results of Testing for Biotinidase Deficiency

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screening calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary 2nd Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Alaska	9,866	9,821	99.54%			7,759	78.64%	215	2.18%
2 Arizona	85,470	81,956	95.89%			57,352	67.10%		
3 Colorado	65,679	63,219	96.25%	60,530	92.16%				
4 Connecticut	43,370	43,727	* 100.82%					191	0.44%
5 Hawaii	17,638	17,612	99.85%					186	1.05%
6 Idaho	19,863	20,254	* 101.97%			13,804	69.50%	199	1.00%
7 Illinois	181,984	188,164	* 103.40%						
8 Indiana	87,891	37,450	42.61%					13,190	15.01%
9 Louisiana	68,275	67,843	99.37%						
10 Maine	13,462	13,341	99.10%						
11 Maryland	69,574	72,390	* 104.05%	61,152	87.89%				
12 Massachusetts	82,673	82,703	* 100.04%					8,818	10.67%
13 Michigan	134,889	134,022	99.36%						
14 Nebraska	24,961	24,863	99.61%						
15 Nevada	30,387	30,659	* 100.90%	25,668	84.47%				
16 New Mexico	26,809	25,745	96.03%	23,581	87.96%			168	0.63%
17 New York	259,995	258,449	99.41%						
18 Oregon	46,790	46,879	* 100.19%	43,423	92.80%			862	1.84%
19 Rhode Island	13,180	13,150	99.77%					427	3.24%
20 Vermont	6,277	6,040	96.22%					326	5.19%
21 Virginia	96,755	99,410	* 102.74%					12,667	13.09%
22 Wisconsin	68,250	66,614	97.60%			1,196	1.75%	5	0.01%
23 Wyoming	5,847	5,480	93.72%						
24 NeoGen Screening	144,319	140,789	97.55%			1,984	1.37%	1,546	1.07%
TOTAL	a 1,604,204	1,550,580	b	214,354	b	82,095	b	38,800	b

* Percentage > 100% denotes inability of program to separate newborns screened from specimens received; **a** = totals should be viewed as a rough estimate due to inability of many states to know actual number of Newborns screened. Many programs count samples received but cannot eliminate duplication when multiple samples are received; **b** = cannot be calculated due to inaccurate data.

9. Congenital Adrenal Hyperplasia (CAH)

9.0 Introduction

Screening for congenital adrenal hyperplasia (CAH) has only been included in U.S. screening programs since 1987. The screening technique used is to determine the concentration of 17-hydroxyprogesterone (17-OHP) in order to detect CAH due to 21-hydroxylase deficiency, which accounts for over 90% of CAH cases. The primary importance of newborn screening is to detect those cases of CAH that might lead to early death due to an unrecognized salt-wasting crisis, or cases of severe virilization in females that might lead to missed sex assignments. In addition, programs detect other types of CAH that may be manifested through short stature due to advanced bone aging, fertility problems in females, and early puberty in males.

The principal laboratory methodology has been radioimmunoassay, although various other types of non-isotopic immunoassay techniques are becoming available. Screening cutoff levels vary among programs, especially as they relate to premature or low birth weight newborns, the source of the majority of false positive results. All data reported here was collected from questionnaires submitted to programs as noted in Chapter 3. Only programs with CAH screening included in their testing scheme are listed in the Tables.

9.1 Definitions for Classical Salt Wasting CAH

Because CAH is the term used for a group of disorders, it is important to know as much about the definitions used in the screening program as possible. An arbitrary sub-classification system for 21-OH CAH has arisen over the years and those classifications are used in many screening programs. In order to ensure that data are correctly related to program definitions, programs were asked to give the definition used for the classical salt wasting type of 21-OH CAH for tabulation in Table 9.01. The question that persists in these definitions is the role that elevated renin levels play in defining salt wasting absent low sodium and high potassium levels.

9.2 Definitions for Classical Simple Virilizing CAH

Programs were asked to give the definition used to define classical simple virilizing CAH and the responses are listed in Table 9.02. The major factor to be considered here is how to differentiate between classical simple virilizing CAH and the less severe nonclassical type, especially in asymptomatic males. This question becomes moderately important in deciding on treatment and follow-up regimens in order to prevent undesirable outcome.

9.3 Definitions for Non-Classical CAH

Programs were asked to give their definition for non-classical CAH and their responses are listed in Table 9.03. It is of particular importance to see how the differentiation between non-classical and classical simple virilizing CAH are made, especially in asymptomatic males.

9.4 Definitions for Unclassified Types of CAH

Because not all cases of CAH detected through screening cannot be included in the first three definitions given in Sections 9.1 - 9.3, programs were asked to define other types of CAH and the responses are given in Table 9.04.

9.5 Laboratory Procedures

In order to identify laboratory procedures in use, programs were asked to report their laboratory protocols and definitions for samples considered 'not normal.' These data are reported in Table 9.05.

9.6 Initial Screening Results

In order to ascertain the effectiveness of screening for CAH, programs were asked to report their findings on initial screening. Initial screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of newborns screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated. Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 9.06. Programs wishing to explain some of their responses included information as footnotes to the table. Because programs continue to report inaccurate or inappropriate data in some of their responses, no totals were given in some columns of the table. Some care should be taken in using these data, since not all programs reported data.

9.7 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are reported in Table 9.07 and are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when a second test is performed without regard for the initial screening result. Programs that requested a second screen because of either a certain result on the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as 'repeat screens' and these are also included. For each case, the data requested included the total newborns tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. It should be recognized that this is a different way of reporting these data than previously and may, therefore, be somewhat confusing. It is hoped that these data better represent the findings from additional screens. Once again, the reader is cautioned about using the data without regard to notes of explanation.

9.8 Cases of Classical Salt Wasting CAH Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of classical salt wasting CAH, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 9.08. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

9.9 Cases of Classical Simple Virilizing CAH Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of classical simple virilizing CAH, whether detected on first or second screen, divided by sex and race/ethnicity as in Section 9.8. The results of this question are given in Table 9.09.

9.10 Cases of Non-Classical CAH Divided by Sex and Race/Ethnicity

Programs were asked to report all cases of non-classical CAH as in Sections 9.8 and 9.9. The results of this question are given in Table 9.10.

9.11 Cases of Unclassified Types of CAH Divided by Sex and Race/Ethnicity

Programs were asked to report all cases of CAH that could not be classified using the definitions in place for the various defined types of CAH and these results are given in Table 9.11.

9.12 Cases of Other Than 21-OH CAH Divided by Sex and Race/Ethnicity

Because some programs have reported cases of CAH that were found not to result from 21-OH deficiency, these data were also requested divided by sex and race/ethnicity. The data are displayed in Table 9.12.

9.13 Time Until Treatment for Classical Salt Wasting CAH

In order to look at overall program efficiency with respect to the most life-threatening type of CAH, programs were asked to report the number of days from birth until treatment of classical salt wasting CAH. The general definition of treatment used in the questionnaire was “initiation of dietary or drug regimen” and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 9.13. In this particular disorder, it is essential that identification of the suspected salt waster be relayed to the treating physician quickly since salt wasting crises can occur within the first week of life and, if unrecognized, can result in death.

9.14 Time Until Treatment for Non-Salt Wasting CAH

Programs were asked to report the time until treatment for the non-salt wasting form of CAH since this type may be more difficult to recognize and is also not usually life threatening. Readers are encouraged not to rush to false conclusions about treatment in these cases since the non-classical type of CAH may go untreated (but not necessarily unrecognized) by consultant specialists. The reported data are listed in Table 9.14.

9.15 Historic Data

In order to document the value of screening for CAH, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of cases detected over the years. These data are given in Table 9.15. Many programs are still wrestling with the complexities of CAH diagnoses and so the case detection data in this table should be used with care.

9.16 Total Newborns Screened

Table 9.16 gives a summation of the data contained in Tables 9.06 and 9.07 along with the number of births in each state or territory as reported in Table 1.01. This table was included so that a tabulation of these data could be viewed without having to refer to several tables.

9.17 Summation of Results

Table 9.17 sums the significant testing data from this Chapter. It includes the total births from Table 1.01, the total newborns reported as being screened, and the testing data reported in Tables 9.06 and 9.07. Comparative percentages have been provided at the request of a number of readers, but in many cases these data are based on estimates or on data that appear to contain duplicate information, thus total compliance percentages may exceed 100% in some cases. Data responders are requested to review these data and adjust their responses to future questionnaires so that more meaning can be associated with these data. In particular, the data in this table seem to point out the difficulty of many programs in separating total samples received from total births screened (i.e. many cannot differentiate between first tests received and others that might have been submitted as independent second tests or repeat screens).

Table 9.01: Definitions for Classical Salt Wasting Congenital Adrenal Hyperplasia

State/Territory	Definition for Classical Salt Wasting Congenital Adrenal Hyperplasia
1 Alabama	NR
2 Alaska	Clinical presentation compatible with 21-OH deficiency, presenting in neonatal period (birth-6 weeks), with confirmatory testing on serum 17-OH progesterone > 3,000 ng/dL, plus salt wasting and elevated plasma.
3 Colorado	Diagnosis made by pediatric endocrinologist.
4 Connecticut	Classified by treatment center.
5 Florida	Persistently elevated 17-OHP and ACTH serum concentrations supported by symptoms (salt wasting) or confirmatory diagnostic lab test.
6 Georgia	Extremely high 17-OHP, normal sodium and potassium. Confirmed by endocrinologist.
7 Hawaii	Clinical presentation compatible with 21-OH deficiency, presenting in neonatal period, with confirmatory serum testing with serum 17-OHP >3000 ng/mL, plus salt wasting (low Na).
8 Illinois	Diagnosis and classification is determined by pediatric endocrinologist.
9 Indiana	Elevated 17-OHP. Diagnosis is made by medical consultant using diagnostic testing.
10 Iowa	High levels of 17-OH progesterone and abnormal electrolytes.
11 Maine	Elevated 17-OHP. Diagnosis is made by medical consultant.
12 Massachusetts	Elevated 17-OHP. Diagnosis is made by medical consultant.
13 Michigan	21-hydroxylase deficiency with salt-wasting.
14 Minnesota	17-OHP > 80 ng/dL. Classification determined by pediatric endocrinologist.
15 New Mexico	Persistently elevated 17-OHP, supported by salt-wasting symptoms and confirmatory diagnostic testing.
16 North Carolina	Elevated 17-OHP, diagnosis and classification made by medical consultant.
17 North Dakota	Persistently elevated 17-OHP supported by symptoms and/or confirmatory diagnostic test (abnormal electrolytes).
18 Rhode Island	Definition and classification determined by endocrine consultant.
19 South Carolina	Determined by specialist.
20 Tennessee	Defined by individual endocrinologist.
21 Texas	Virilized genitalia; serum 17-OHP >10,000 ng/dL; Na <130; K >7; urinary Na/K ratio >2.5; Advanced bone age.
22 Washington	Persistently elevated 17-OHP supported by salt-wasting symptoms and confirmatory diagnostic testing.
23 Wisconsin	Diagnosis and classification is made by treatment center.
24 NeoGen Screening	Classification by an endocrinologist.

Table 9.02: Definitions for Classical Simple Virilizing Congenital Adrenal Hyperplasia

State/Territory	Definition for Classical Simple Virilizing Congenital Adrenal Hyperplasia
1 Alabama	NR
2 Alaska	Clinical presentation compatible with 21-OH deficiency, presenting in neonatal period (birth-6 weeks), or later childhood in males with confirmatory serum 17-OH progesterone > 3,000ng/mL and no evidence of salt wasting.
3 Colorado	Diagnosis made by pediatric endocrinologist.
4 Connecticut	Classified by treatment center.
5 Florida	Patients diagnosed with CAH who have manifestations of advanced bone ages, genital ambiguity, elevated serum concentrations of 17-OHP and ACTH, but normal levels of sodium, potassium, and plasma renin activity.
6 Georgia	Lower, but elevated 17-OHP, normal sodium and potassium, often not detected at 1st test but gradually escalates, occasionally females are virilized.
7 Hawaii	Clinical presentation compatible with 21-OH deficiency, presenting in neonatal period, or later childhood with confirmatory serum 17-OHP > 3000 mg/mL, no evidence of salt-wasting.
8 Illinois	Diagnosis and classification is determined by pediatric endocrinologist.
9 Indiana	Elevated 17-OHP. Diagnosis is made by medical consultant using diagnostic testing.
10 Iowa	High levels of 17-OH progesterone, normal electrolytes.
11 Maine	Elevated 17-OHP. Diagnosis is made by medical consultant.
12 Massachusetts	Elevated 17-OHP. Diagnosis is made by medical consultant.
13 Michigan	21 hydroxylase without salt-wasting.
14 Minnesota	Final classification determined by pediatric endocrinologist.
15 New Mexico	Elevated 17-OHP. Diagnosis made by medical consultant.
16 North Carolina	Elevated 17-OHP, diagnosis and classification made by medical consultant.
17 North Dakota	High levels of 17-OH progesterone, normal electrolytes.
18 Rhode Island	Definition and classification determined by endocrine consultant.
19 South Carolina	Determined by specialist.
20 Tennessee	Defined by individual endocrinologist.
21 Texas	Virilized genitalia serum; 17-OHP >10,000 ng/dL; Na >130; K <7; Urinary Na/K ratio <2.5; Advancing bone age.
22 Washington	Persistently elevated 17-OHP without salt-wasting symptoms supported by confirmatory diagnostic testing.
23 Wisconsin	Diagnosis and classification is made by treatment center.
24 NeoGen Screening	Classification by an endocrinologist.

Table 9.03: Definitions for Non-Classical Congenital Adrenal Hyperplasia

State/Territory	Definition for Non-Classical Congenital Adrenal Hyperplasia
1 Alabama	NR
2 Alaska	Compatible clinical features w/basal serum 17-OH-P <2,000ng/dL but an abnormal increase after ACTH stimulation > 2,000ng/mL.
3 Colorado	Diagnosis made by pediatric endocrinologist.
4 Connecticut	Classified by treatment center.
5 Florida	The mildist form of CAH characterized by persistent slight elevations of 17-OHP.
6 Georgia	NR
7 Hawaii	Compatible clinical features w/basal serum 17-OH-P <2,000ng/dL but an abnormal increase after ACTH stimulation > 2,000ng/mL.
8 Illinois	Diagnosis and classification is determined by pediatric endocrinologist.
9 Indiana	Elevated 17-OHP. Diagnosis is made by medical consultant using diagnostic testing.
10 Iowa	High levels of stimulated 17-OH progesterone usually detected at a later age.
11 Maine	Elevated 17-OHP. Diagnosis is made by medical consultant.
12 Massachusetts	Elevated 17-OHP. Diagnosis is made by medical consultant.
13 Michigan	21 hydroxylase deficiency without salt-wasting presenting after infancy.
14 Minnesota	Final classification determined by pediatric endocrinologist.
15 New Mexico	Elevated 17-OHP. Diagnosis made by medical consultant.
16 North Carolina	Diagnosis and classification made by medical consultant.
17 North Dakota	High levels of stimulated 17-OH progesterone usually detected at a later age.
18 Rhode Island	Definition and classification determined by endocrine consultant.
19 South Carolina	Determined by specialist.
20 Tennessee	Defined by individual endocrinologist.
21 Texas	Normal genitalia; 17-OHP 500-10,000 ng/dL; Normal urinary Na/K ratio, normal 11 Doc, and normal bone age.
22 Washington	CAH due to an enzyme deficiency other than 21-hydroxylase.
23 Wisconsin	Diagnosis and classification is made by treatment center.
24 NeoGen Screening	Classification by an endocrinologist.

Table 9.04: Definitions for Unclassified Congenital Adrenal Hyperplasia

State/Territory	Definition for Unclassified Congenital Adrenal Hyperplasia
1 Alabama	High 17-OHP and follow-up by physician.
2 Alaska	N/R
3 Colorado	Diagnosis made by pediatric endocrinologist.
4 Connecticut	Classified by treatment center.
5 Florida	Patients diagnosed with CAH for whom a diagnostic classification has not been established.
6 Georgia	NR
7 Hawaii	NR
8 Illinois	Diagnosis and classification is determined by pediatric endocrinologist.
9 Indiana	Elevated 17-OHP. Diagnosis is made by medical consultant using diagnostic testing.
10 Iowa	High levels of stimulated adrenal steroid precursors other than 17-OH progesterone.
11 Maine	NR
12 Massachusetts	Elevated 17-OHP. Diagnosis is made by medical consultant.
13 Michigan	21 hydroxylase deficiency and status regarding salt-wasting is unknown.
14 Minnesota	Final classification determined by pediatric endocrinologist.
15 New Mexico	NA
16 North Carolina	Diagnosis and classification made by medical consultant.
17 North Dakota	High levels of stimulated adrenal steroid precursors other than 17-OH progesterone.
18 Rhode Island	Definition and classification determined by endocrine consultant.
19 South Carolina	Case records that do not specify type of CAH.
20 Tennessee	Defined by individual endocrinologist.
21 Texas	17-OHP > 500 ng/dL; uncertain of defect.
22 Washington	Persistently elevated 17-OHP without symptoms or supporting diagnostic evidence.
23 Wisconsin	Diagnosis and classification is made by treatment center.
24 NeoGen Screening	Classification by an endocrinologist.

Table 9.05: Congenital Adrenal Hyperplasia - Laboratory Testing

State/Territory	Testing Method	Definition of Tiers	Definition of NOT NORMAL (ng/mL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (ng/mL) <i>Requiring Immediate Serum Follow-up</i>
1 Alabama	Radioimmunoassay	> 24hrs.old (> 2500g) < 48 hrs. old (> 2500g) Low birth weight (≤ 2500g) (premature births)		25-45 ng/mL, 45-65 ng/mL, 45-65 ng/mL.
2 Alaska	RIA	0-10 days (0-35µg/mL) >10 days (0-25µg/ml) LBW(<5.5lbs)0-45µg/mL	> 35ng/mL (0-10 days) > 25ng/mL (>10 days) > 45 ng/mL (LBW < 5.5)	> 80 ng/mL.
3 Colorado	NR	NR	NR	NR
4 Connecticut	FIA	BW < 1700 Grams BW > 1700 Grams	115-134 ng/mL 1/1/00 to 3/31/00 130-150 ng/ml 4/1/00 to 12/31/00 53-89 ng/mL 1/1/00 to 3/31/00 65-100 ng/mL 4/1/00 to 12/31/00	≥ 135 ng/mL 1/1/00 to 3/31/00 ≥ 150 ng/mL 4/1/00 to 12/31/00 ≥ 90 ng/mL 1/1/00 to 3/31/00 ≥ 100 ng/mL 4/1/00 to 12/31/00
5 Florida	TRIF	< 1999g borderline ≥ 2000g < 12 hrs Borderline > 12 hrs Borderline	150-224 71-149 41-89	≥ 225 ≥ 150 ≥ 90
6 Georgia	RIA (17-OHP)	< 2500 gm ≥ 2500 gm	80-199 < 48 hrs; 80-149 ≥ 48 hrs. 80-119 < 48 hrs; 45-99 ≥ 48 hrs.	≥ 200ng/mL <48 hrs; ≥ 150ng/mL >48 hrs. >120 ng/mL <48 hrs; ≥ 100 ≥ 48 hrs.
7 Hawaii	RIA	> 5 lbs 8 oz. ≤ 5 lbs 8 oz.	> 35 ng/mL < 10 days; > 25 ng/mL > 10 days > 60 ng/mL < 10 days; > 40 ng/mL > 10 days	> 80 ng/mL < 10 days; > 50 ng/mL >10 days > 120 ng/mL < 10 days; > 50 ng/mL > 10 days
8 Illinois	FIA	36 weeks gest. or > 2500 grams 36 weeks gest. or < 2500 grams	≥ 54 ng/mL; < 89 ng/mL ≥ 90 ng/mL; < 129 ng/mL	≥ 90 ≥ 130
9 Indiana	FIA	> 2500 gm 1500 - 2499 gm < 1500 gm	50-80 65-130 80-150	> 80 > 130 ≥ 150
10 Iowa	FIA FIA	< 1250 g ≥ 1250 < 1750 g ≥ 1750 < 2250 g ≥ 2250 g	≥ 135 < 160 ≥ 90 < 135 ≥ 65 < 90 ≥ 50 but < 90	≥ 160 ≥ 135 ≥ 90 ≥ 90
11 Maine	FIA	> 1500 g; ≤ 1500 g	≥ 50 ng/mL; ≥ 50 ng/mL.	
12 Massachusetts	FIA	0-1000g 1001-1750g 1751-2250g > 2250g	≥ 100 ng/mL ≥ 80 ng/mL ≥ 50 ng/mL ≥ 50 ng/mL	≥ 135 ng/mL ≥ 100 ng/mL ≥ 70 ng/mL ≥ 50 ng/mL
13 Michigan	FIA	> 2500 NBW ≤ 2500 LBW	100-149 ng/mL ≤ 12 hrs old; 55-74 ng/mL > 12 hrs old. 100 - 149 ng/mL any age	≥ 150 ng/mL ≤ 12 hrs old; 75 ng/mL > 12 hrs old. ≥ 150 ng/mL any age
14 Minnesota	FIA	< 1500 g 1500-2500 g ≥ 2500 g	80-150 ng/mL 65-130 ng/mL 50-80 ng/mL	> 150 ng/mL > 130 ng/mL > 80 ng/mL
15 New Mexico	Delphia	< 1250 gm 135-159 Borderline Positive > 160 ng/mL 1751-2249 gm > 90 ng/mL-pos > 2200 gm > 90 ng/mL - positive	Borderline 50 - 89	90 and above
16 North Carolina	Delphia Fluro-immunoassay	< 1399 g 1400-1799 g ≥ 1800 g	≥ 130 ≥ 100 ≥ 60	≥ 130 ≥ 100
17 North Dakota	FIA	<1250 g ≥ 1250 < 1750 g ≥ 1750 < 2250 g ≥ 2250 g	≥ 135 ng/mL < 160 ≥ 90 < 135 ≥ 65 < 90 ≥ 50 < 90 ng/mL.	≥ 160 ≥ 135 ≥ 90 ≥ 90
18 Rhode Island	FIA	0-1000 g 1000-1750 g 1751-2250 g > 2250 g	< 100 ng/mL < 80 ng/mL < 50 mg/mL < 50 ng/mL	≥ 150 - 200 ng/mL
19 South Carolina	EIA	> 2500 grams a ≤ 2500 grams	≥ 40ng/mL ≥ 65ng/mL	DA DA
20 Tennessee	FIA	<1250 gms 1250 - 1749 gms 1750 -2249 gms ≥ 2250 gms	135 - 159 ng/mL 90 - 134 ng/mL 65 - 89 ng/mL 50 - 89 ng/mL	≥ 160 ng/mL ≥ 135 ng/mL ≥ 90 ng/mL ≥ 90 ng/mL
21 Texas	RIA-17OHP	< 2500 grams - low birth weight > 2500 grams - normal birth wt.	> 99 ng/mL > 45 ng/mL	≥ 400 ng/mL ≥ 100 ng/mL after 2nd screen, low Na, high K, ambiguous genitalia
22 Washington	RIA	≤ 24 hrs or ≤ 2500 g ≥ 24 hrs or ≥ 2500 g	≥ 71 ng/mL ≥ 43 ng/mL	≥ 100 ng/mL
23 Wisconsin	Delfia	≤ 1299 g 1300-1699 g 1700-2199 g ≥ 2200 g	requires immediate filter paper testing ≥ 135 115-134 ≥ 135 65-89 ≥ 90 55-89 ≥ 90	b
24 NeoGen Screening	1) Immunofluorescent assay 2) Ether Extraction	1) BW = >3000g, 2500-3000g 1500-2500g, <1500g 17OHP=<17.3, <22.7, <27.3, <45.5 2) Ether Extraction cutoff < 15.0 mg/dL	extracted 17-OHP cutoff ≥ 15.0 ng/mL	for infants ≥ 3000 g the critical cutoff for the total 17-OHP is 68 ng/mL or when a second sample 17-OHP is greater than the first sample 17-OHP

LBW = Low birth weight

NBW = Normal birth weight

a = all results where the 17-OHP is ≥ 100 for infants > 2500 g BW and ≥ 130 for infants ≤ 2500 g BW are called to the physician of record; b = serum follow-up not recommended based on initial filter paper result.

Table 9.06: Initial Screening Results - Congenital Adrenal Hyperplasia

State/Territory	Number Newborns Screened	Number Newborns with <u>NOT NORMAL</u> Test Results	Number Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number Newborns Confirmed with Classical CAH Salt Wasting	Number Newborns Confirmed with Classical CAH Simple Virilizing	Number Newborns Confirmed with Non-Classical CAH	Number Newborns Confirmed with 21-OH deficient CAH unable to classify	Number Newborns with other than 21-OH deficiency		
								11-OH	3β - HSD	Other
1 Alabama	62,173						3			
2 Alaska	9,821	133	0	2	0	0	0	0	0	0
3 Colorado	63,219	540	9 i	0	DA	1	DA			
4 Connecticut	43,730	78	0	2	2	0	0			
5 Florida	204,030	867 a	64 b	6	2	3	1	0	0	0
6 Georgia	189,498			5	1					
7 Hawaii	17,612	74	11 j	0	0	0	0			
8 Illinois	188,164	1,068	0	8	3	0	0	0	0	0
9 Indiana	21,647	139	1	0	1	0	0	0	0	0
10 Iowa	38,141	66	9 k	2	1	0	0	0	0	0
11 Maine	13,341	70	0	0	0	0	0	0	0	0
12 Massachusetts	82,703	757	N/A	5	2		4			
13 Michigan	134,022	794		11	1					
14 Minnesota	68,402	66		4	1		1			
15 New Mexico	25,745	232		1	NA	NA	1			
16 North Carolina	120,750	481	N/D	6	NA	NA	NA	N/C	N/C	N/C
17 North Dakota	8,806	65	1 d	1	0	0	0	0	0	0
18 Rhode Island	13,150									
19 South Carolina	52,176	400	16	1 c	0	0	1 h	0	0	0
20 Tennessee	79,539	115		1						
21 Texas	355,100			17 l	7 m	0	4			1
22 Washington	76,886	77	0	4	0	0	0			
23 Wisconsin	66,614	311	5	2	0	0	0			
24 NeoGen Screening	140,789	268	25 e	1	1	1	3			
TOTAL	f	f	f	79 g	22 g	5 g	18 g	0 g	0 g	1 g

a = includes 801 borderline, 66 presumptive abnormal, 4 confirmed CAH were borderline, borderline followed for repeat screen only, letter sent to parent if repeat not received within 21 days; b = all lost to follow-up were borderline, closed at 180 days; c = had ambiguous genitalia, initial specimen unsat; d = deceased; e = physician notification; f = totals not given - many programs reported inaccurate or inappropriate data; g = total given does not include all states; h = premature infant, first screen WNL; i = certified letter to mother, not further information; j = expired; k = 4 deceased, 5 parents refused second test; l = one salt wasting case had 2 normal but had a prenatal diagnosis; m = two simple virilizing cases had 2 normal screens and diagnosed later.

Table 9.07: Second Screens for Congenital Adrenal Hyperplasia

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed with Classical CAH Salt Wasting				Number of Newborns Confirmed with Classical CAH Simple Virilizing				Number of Newborns Confirmed with Non-Classical CAH							
	R	D	P		R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total				
1 Alabama		62,731	639	63,370																								
2 Alaska		7,759	215	7,974		23	2	25																				
3 Colorado	60,530			60,530	10			10													1			1				
4 Connecticut			273	273			3	3																				
5 Florida			N/A	0		N/A	N/A	0		N/A	N/A			N/A	N/A			N/A	N/A			N/A	N/A	0				
6 Georgia																												
7 Hawaii			249	249			5	5																				
8 Illinois			N/C	0										0	0													
9 Indiana			7,624	7,624			16	16																				
10 Iowa		DA	1,439	1,439		DA		0		DA	0	0		DA	0	0		DA	0	0		DA	0	0				
11 Maine																												
12 Massachusetts			8,818	8,818			50	50				0				0				0				0				
13 Michigan			0	0				0																				
14 Minnesota			906	906			368	368																				
15 New Mexico	23,531		168	23,699	5			5																				
16 North Carolina		N/C	N/C			8	N/C	8		N/D	N/D			N/A	N/A			N/A	N/A			N/A	N/A					
17 North Dakota		DA	494	494		DA		0		DA	0	0		DA	0	0		DA	0	0		DA	0	0				
18 Rhode Island			427	427																								
19 South Carolina		N/A	3,657	3,657		N/A	N/A			N/A	N/A			0	0	0		0	0	0		0	0	0				
20 Tennessee																												
21 Texas	348,132		1,919	350,051				0				0				0	2			2		8		8				
22 Washington		69,534	1,222	70,756		7	NR	7		0	NR	0			NR	0		1	NR	1		0	NR	0				
23 Wisconsin		1,196	1,424	2,620		1	22	23																				
24 NeoGen Screening		1,984	1,546	3,530	a			0																				
TOTAL	b	432,193	143,204	31,020	c			c				c		0	0	0	0	d	2	1	0	3	d	9	0	0	9	d

a = these numbers are overall; **b** = totals given reflect program responses and should be viewed as estimates only, given the number not reporting and the number of caveats listed;

c = totals not given - too many programs reported inaccurate or inappropriate data; **d** = totals given does not include all states.

continued

Table 9.07: Second Screens for Congenital Adrenal Hyperplasia (continued)

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns Confirmed with 21-OH deficient CAH unable to classify				Number of Newborns with other than 21-OH deficiency																
	R	D	P		R	D	P	Total	11-OH				3B-HSD				Other								
									R	D	P	Total	R	D	P	Total	R	D	P	Total					
1 Alabama		62,731	639	63,370																					
2 Alaska		7,759	215	7,974																					
3 Colorado	60,530			60,530																					
4 Connecticut			273	273																					
5 Florida			N/A	0		N/A	N/A			N/A	N/A			N/A	N/A			N/A	N/A						
6 Georgia																									
7 Hawaii			249	249																					
8 Illinois			N/C	0																					
9 Indiana			7,624	7,624																					
10 Iowa		D/A	1,439	1,439		DA	0	0		DA	0	0		DA	0	0		DA	0	0					
11 Maine																									
12 Massachusetts			8,818	8,818				0	0			0				0				0					
13 Michigan				0																					
14 Minnesota			906	906																					
15 New Mexico	23,531		168	23,699																					
16 North Carolina		N/C	N/C	N/C			N/A	0		N/C	N/C			N/C	N/C			N/C	N/C						
17 North Dakota		D/A	494	494		DA	0	0		DA	0	0		DA	0	0		DA	0	0					
18 Rhode Island			427	427																					
19 South Carolina		N/A	3,657	3,657		0	0	0		0	0	0		0	0	0		0	0	0					
20 Tennessee																									
21 Texas	348,132		1,919	350,051	5		0	5				0				0	1			1					
22 Washington		69,534	1,222	70,756			NR	0			NR				NR				NR						
23 Wisconsin		1,196	1,424	2,620																					
24 NeoGen Screening		1,984	1,546	3,530	a																				
Total	b	432,193	143,204	31,020	c	5	0	0	5	d	0	0	0	0	d	0	0	0	0	d	1	0	0	1	d

a = these numbers are overall; b = totals given reflect program responses and should be viewed as estimates only given, the number not reporting and the number of caveats listed;

c = totals not given - too many programs reported inaccurate or inappropriate data; d = total given does not include all states.

Table 9.08: Cases of Classical Salt Wasting Congenital Adrenal Hyperplasia

Total detected on initial and second screen

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Alabama																	
2 Alaska													0	0	2	¹		
3 Colorado																		
4 Connecticut	1									1		2	0	2				
5 Florida	2	2	1			1						3	3	6				0
6 Georgia	1	2	1									2	2	5	³	1		1
7 Hawaii																		
8 Illinois	5	2								1		6	2	8				
9 Indiana																		
10 Iowa	2											2	0	2				
11 Maine																		
12 Massachusetts											4	1	4	1	5			
13 Michigan	8	3										8	3	11				
14 Minnesota	1	3										1	3	4				
15 New Mexico														1	¹			
16 North Carolina														6	¹			
17 North Dakota	1											1	0	1				
18 Rhode Island																		
19 South Carolina		1												0	1	1		
20 Tennessee				1										0	1	1		
21 Texas	3	1							1	2		4	3	17	³	5	5	10
22 Washington	1	3										1	3	4				
23 Wisconsin	1	1										1	1	2				
24 NeoGen Screening	1											1	0	1				
TOTAL	27	18	2	1	0	1	0	0	1	2	6	1	36	23	79	6	5	11

See footnotes at bottom of Table 9.09

Table 9.09: Cases of Classical Simple Virilizing Congenital Adrenal Hyperplasia

Total detected on initial and second screen

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Alabama															0		
2 Alaska															0			
3 Colorado															0			
4 Connecticut		2										0	2	2		1		1
5 Florida		2										0	2	2				0
6 Georgia												0	0	1	³	1		1
7 Hawaii																		
8 Illinois	1										2	3	0	3				
9 Indiana														1	³	1		1
10 Iowa	1											1	0	1				
11 Maine																		
12 Massachusetts											1	1	1	1	2			
13 Michigan	1											1	0	1				
14 Minnesota		1										0	1	1				
15 New Mexico																		
16 North Carolina															N/A			
17 North Dakota																		
18 Rhode Island																		
19 South Carolina																		
20 Tennessee																		
21 Texas	1	2										1	2	9	³	4	2	6
22 Washington	1											1	0	1				
23 Wisconsin																		
24 NeoGen Screening		1										0	1	1				
TOTAL	5	8	0	0	0	0	0	0	0	0	3	1	8	9	25	4	5	9

¹ did not report a breakdown of Race/Ethnicity.

² total reported is from Table 9.06 and Table 9.07; therefore total does not equal or may differ from data given in Table 9.08 & 9.09 and denotes a problem in reliability.

³ Ethnicity counted as a Race and is included in total.

Table 9.10: Cases of Non-Classical Congenital Adrenal Hyperplasia

Total detected on initial and second screen

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Total	Hispanic		Total
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female		Male	Female	
	1 Alabama																	
2 Alaska																		
3 Colorado											2		2	0	2			
4 Connecticut																		
5 Florida	1	2											1	2	3			0
6 Georgia																		
7 Hawaii																		
8 Illinois																		
9 Indiana																		
10 Iowa																		
11 Maine																		
12 Massachusetts																		
13 Michigan																		
14 Minnesota																		
15 New Mexico																		
16 North Carolina																N/A		
17 North Dakota																		
18 Rhode Island																		
19 South Carolina																		
20 Tennessee																		
21 Texas	1												1	0	8 ³	5	2	7
22 Washington																		
23 Wisconsin																		
24 NeoGen Screening				1									0	1	1			
TOTAL	2	2	0	1	0	0	0	0	0	0	0	0	4	3	14	5	2	7

See footnotes at bottom of Table 9.11

Table 9.11 Cases of Unclassified Congenital Adrenal Hyperplasia

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Total	Hispanic		Total
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female		Male	Female	
	1 Alabama														3 ¹			
2 Alaska																		
3 Colorado																		
4 Connecticut																		
5 Florida				1									0	1	1			
6 Georgia																		
7 Hawaii																		
8 Illinois																		
9 Indiana																		
10 Iowa																		
11 Maine																		
12 Massachusetts											4		4	0	4			
13 Michigan																		
14 Minnesota	1												1	0	1			
15 New Mexico															1 ¹			
16 North Carolina																N/A		
17 North Dakota																		
18 Rhode Island																		
19 South Carolina	1												1	0	1			
20 Tennessee																		
21 Texas	1	2											1	2	9 ³	3	3	6
22 Washington																		
23 Wisconsin																		
24 NeoGen Screening	1		1										2	0	3 ^a			
TOTAL	4	2	1	1	0	0	0	0	0	0	4	0	9	3	23	3	3	6

¹ did not report a breakdown of Race/Ethnicity.

² total reported is from Table 9.06 and Table 9.07; therefore total does not equal or may differ from data given in Table 9.10 & 9.11 and denotes a problem in reliability.

³ Ethnicity counted as a Race and is included in total.

a = one unknown sex.

Table 9.12: Cases of Other Than 21-OH Congenital Adrenal Hyperplasia
 Total detected on initial and second screen

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic				
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total	
1 Alabama																			
2 Alaska																			
3 Colorado																			
4 Connecticut																			
5 Florida																			
6 Georgia																			
7 Hawaii																			
8 Illinois																			
9 Indiana																			
10 Iowa																			
11 Maine																			
12 Massachusetts																			
13 Michigan																			
14 Minnesota																			
15 New Mexico																	NC		
16 North Carolina																	NC		
17 North Dakota																			
18 Rhode Island																			
19 South Carolina																			
20 Tennessee																			
21 Texas	1												1	0	2	³	1		1
22 Washington																			
23 Wisconsin																			
24 NeoGen Screening																			
TOTAL	1	0	0	0	0	0	0	0	0	0	0	0	1	0	2		1	0	1

¹Ethnicity counted as a Race and is included in total

²total reported is from Table 9.06 and Table 9.07; therefore total does not equal or may differ from data given in Table 9.12 and denotes a problem in reliability.

Table 9.13: Days from Birth Until Treatment Initiated for Classical Salt Wasting Congenital Adrenal Hyperplasia
Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	Total
1 Alabama																
2 Alaska																2 ¹
3 Colorado																0
4 Connecticut							1		1							2
5 Florida	1		1			1	2	1								6
6 Georgia	2								1				2			5
7 Hawaii																
8 Illinois	1			1		2		1	1			1		1		8
9 Indiana																
10 Iowa				1	1											2
11 Maine																
12 Massachusetts															5	5
13 Michigan															11	11
14 Minnesota	3						1									4
15 New Mexico				1												1
16 North Carolina															6	6
17 North Dakota					1											1
18 Rhode Island																
19 South Carolina													1			1
20 Tennessee															1	1
21 Texas	7							2		1	3		1	3		17
22 Washington	2	1			1											4
23 Wisconsin			1									1				2
24 NeoGen Screening															1	1

¹did not report a breakdown of Days from Birth.

²total reported is from Table 9.06 and Table 9.07; therefore total does not equal or may differ from data given in Table 9.13 & 9.14 and denotes a problem in reliability.

Table 9.14: Days from Birth Until Treatment Initiated for Non-Salt Wasting Congenital Adrenal Hyperplasia (Simple Virilizing and Non-Classical)

Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	Total
1 Alabama																
2 Alaska																
3 Colorado														2		2
4 Connecticut	1												1			2
5 Florida				1	1									1	2	5
6 Georgia														1		1
7 Hawaii																
8 Illinois						1		1							1	3
9 Indiana	1															1
10 Iowa						1										1
11 Maine																
12 Massachusetts															2	2
13 Michigan															1	1
14 Minnesota			1													1
15 New Mexico																
16 North Carolina																N/A
17 North Dakota																
18 Rhode Island																
19 South Carolina																
20 Tennessee																
21 Texas													2	3	4	17 ^a
22 Washington															1 ^b	1
23 Wisconsin																0
24 NeoGen Screening															2	2

¹did not report a breakdown of Days from Birth.

²total reported is from Table 9.06 and Table 9.07; therefore total does not equal or may differ from data given in Table 9.13 & 9.14 and denotes a problem in reliability.

a = includes 9 simple virilizing only, did not breakdown non-classical; **b** = false negative on first screen.

Table 9.15: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Classical Salt Wasting CAH			Classical Simple Virilizing CAH			Non Classical CAH			Other Than 21-OH CAH			Unclassified Type of CAH		
	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases
1 Alabama	NR			NR			NR			NR			NR		
2 Alaska	1987	13	12	1987	13	7	1987	13	2			NR			NR
3 Colorado	Aug-00	5 months	0				Aug-00	5 months	2						
4 Connecticut	Oct-97	3 1/4	5	Oct-97	3 1/4	2	Oct-97	3 1/4	0	Oct-97	3 1/4	0	Oct-97	3 1/4	0
5 Florida	Apr-95	5 4/5	30	Apr-95	5 4/5	5	Apr-95	5 4/5	8	Apr-95	5 4/5	1	Apr-95	5 4/5	11
6 Georgia	Jun-90	9 1/2	51	Jun-90	9 1/2	12	Jun-90	9 1/2	0	Jun-90	9 1/2	0	Jun-90	9 1/2	0
7 Hawaii	Jul-97	2 1/2	1	Jul-97	2 1/2	0	Jul-97	2 1/2	0	Jul-97	2 1/2	0	Jul-97	2 1/2	0
8 Illinois	1986	14	74	1986	14	21	NR		NR	NR		NR	NR		NR
9 Indiana	Oct-00	1/4	0	Oct-00	1/4	1	Oct-00	1/4	0	Oct-00	1/4	0	Oct-00	1/4	0
10 Iowa	N/C		N/C	N/C		N/C	N/C		N/C	N/C		N/C	N/C		N/C
11 Maine	1998	2	0												
12 Massachusetts	1995	5	53												
13 Michigan	1993	7	53	1993	7	7									
14 Minnesota	NR														
15 New Mexico	1998	2	2	1998	2	0	1998	2	1	1998	2	1	1998	2	1
16 North Carolina	NC		N/C	N/C		N/C	N/C		N/C	DA		DA	DA		DA
17 North Dakota	1992	8	11												
18 Rhode Island															
19 South Carolina	Oct-92	8yr. 3 mo.	15	Oct-92	8yr. 3 mo.	3	Oct-92	8yr. 3 mo.	1	Oct-92	8yr. 3 mo.	2	Oct-92	8 yr. 3 mo.	7
20 Tennessee	NR														
21 Texas	1989	11	187	1989	11	72	1989	11	103	1989	11	17	1989	11	35
22 Washington	Jul-84	16 1/2	55	Jul-84	16 1/2	11	Jul-84	16 1/2	7	DA		DA	Jul-84	16 1/2	6
23 Wisconsin	1993	7	38	1993	7	4	1993	7	3	NR		NR	1993	7	1
24 NeoGen Screening	NR														

a = includes sv, non-classical, 21-OH, and other than 21-OH.

Table 9.16: Total Newborns Screened for Congenital Adrenal Hyperplasia

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to follow-up	Number of Newborns Confirmed w/Classical CAH Salt Wasting	Number of Newborns Confirmed w/ Classical CAH-Simple Virilizing	Number of Newborns Confirmed w/ Non-Classical CAH	Number of Newborns Confirmed w/ 21-OH deficient CAH unable to Classify	Number of Newborns with other than 21-OH deficiency		
								11-OH	3β-HSD	Other
1 Alabama	62,562		0	0	0		3			
2 Alaska	9,866	158	0	2	0	0	0	0	0	0
3 Colorado	65,679	550	9	0	0	2	0	0	0	0
4 Connecticut	43,370	81	0	2	2	0	0	0	0	0
5 Florida	204,305	867	64	6	2	3	1	0	0	0
6 Georgia	133,524	0	0	5	1	0	0	0	0	0
7 Hawaii	17,638	79	11 a	0	0	0	0	0	0	0
8 Illinois	181,984	1,068	0	8	3	0	0	0	0	0
9 Indiana	87,891	155	1	0	1	0	0	0	0	0
10 Iowa	38,418	66	9	2	1	0	0	0	0	0
11 Maine	13,462	70	0	0	0	0	0	0	0	0
12 Massachusetts	82,673	807	0	5	2	0	4	0	0	0
13 Michigan	134,889	794	0	11	1	0	0	0	0	0
14 Minnesota	67,546	434	0	4	1	0	1	0	0	0
15 New Mexico	26,809	237	0	1	0	0	1	0	0	0
16 North Carolina	121,347	489	ND	6	NA	NA	NA	NC	NC	NC
17 North Dakota	8,847	65	1	1	0	0	0	0	0	0
18 Rhode Island	13,180	0	0	0	0	0	0	0	0	0
19 South Carolina	53,562	400	16	1	0	0	1	0	0	0
20 Tennessee	84,832	115	0	1	0	0	0	0	0	0
21 Texas	368,019	0	0	17	9	8	9	0	0	2
22 Washington	80,453	84	0	4	1	0	0	0	0	0
23 Wisconsin	68,250	334	5	2	0	0	0	0	0	0
24 NeoGen Screening	144,319	268	25	1	1	1	3	0	0	0
TOTAL	2,113,425	7,121	141	79 b	25 b	14 b	23 b	0 b	0 b	2 b

a = expired; **b** = total given does not include all states.

Table 9.17: Summation Results of Testing for Congenital Adrenal Hyperplasia

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screening calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary 2nd Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Alabama	62,562	62,173	99.38%			62,731	100.27%	639	1.02%
2 Alaska	9,866	9,821	99.54%			7,759	78.64%	215	2.18%
3 Colorado	65,679	63,219	96.25%	60,530	92.16%				
4 Connecticut	43,370	43,730	* 100.83%					273	0.63%
5 Florida	204,305	204,030	99.87%						
6 Georgia	133,524	189,498	* 141.92%						
7 Hawaii	17,638	17,612	99.85%					249	1.41%
8 Illinois	181,984	188,164	* 103.40%						
9 Indiana	87,891	21,647	24.63%					7,624	8.67%
10 Iowa	38,418	38,141	99.28%					1,439	3.75%
11 Maine	13,462	13,341	99.10%						
12 Massachusetts	82,673	82,703	* 100.04%					8,818	10.67%
13 Michigan	134,889	134,022	99.36%						
14 Minnesota	67,546	68,402	* 101.27%					906	1.34%
15 New Mexico	26,809	25,745	96.03%	23,531	87.77%			168	0.63%
16 North Carolina	121,347	120,750	99.51%						
17 North Dakota	8,847	8,806	99.54%					494	5.58%
18 Rhode Island	13,180	13,150	99.77%					427	3.24%
19 South Carolina	53,562	52,176	97.41%					3,657	6.83%
20 Tennessee	84,832	79,539	93.76%						
21 Texas	368,019	355,100	96.49%	348,132	94.60%			1,919	0.52%
22 Washington	80,453	76,886	95.57%			69,534	86.43%	1,222	1.52%
23 Wisconsin	68,250	66,614	97.60%			1,196	1.75%	1,424	2.09%
24 NeoGen Screening	144,319	140,789	97.55%			1,984	1.37%	1,546	1.07%
TOTAL	a 2,113,425	2,076,058	b	432,193	b	143,204	b	31,020	b

* Percentage > 100% denotes inability of program to separate infants screened from specimens received; **a** = totals should be viewed as a rough estimate due to inability of many states, to know actual number of infants screened. Many programs count samples received but cannot eliminate duplication when multiple samples are received; **b** = cannot be calculated due to inaccurate data.

10. Cystic Fibrosis (CF)

10.0 Introduction

Screening for cystic fibrosis is included in only a few screening programs. The debate continues as to the benefits of newborn screening for CF given the lack of a curative treatment regimen. It is now generally accepted that a stepwise screening procedure, including screening for immunoreactive trypsin, secondary screening with DNA for $\Delta F508$ (the most frequent and most severe mutation in the U.S. population), and confirmatory sweat testing, provide the most effective screening protocol.

All data reported here were collected from questionnaires submitted to programs as noted in Chapter 3. Only programs with CF screening included in their testing scheme are listed in the Tables. Connecticut is included because the screening program in place at the University of Connecticut Medical School accounts for a significant number of screened newborns even though it is not 'officially' a part of the state screening program.

10.1 Laboratory Procedures

In order to identify laboratory procedures in use, programs were asked to report their laboratory protocols and definitions for samples considered 'not normal.' These data are reported in Table 10.01. Although only a few programs are screening, several different approaches to laboratory testing exist along with varying definitions of 'not normal.' Research continues as to the best approach to screening.

10.2 Initial Screening Results

In order to ascertain the effectiveness of screening for CF, programs were asked to report their findings on initial screening. Initial screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of infants screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated. Additionally,

programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 10.02. Programs wishing to explain some of their responses included information as footnotes to the table. Because some requested data were not submitted, some care should be exercised in their use.

10.3 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are reported in Table 10.03 and are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when a second test is performed without regard for the initial screening result. Programs that requested a second screen because of either a certain result on the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as 'repeat screens' and these are also included.

For each case, the data requested included the total infants tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. It should be recognized that this is a different way of reporting these data than previously and may, therefore, be somewhat confusing. It is hoped that these data better represent the findings from additional screens. Once again, the reader is cautioned about using the data without regard to notes of explanation.

10.4 Cases of CF Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of CF, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 10.04.

For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

10.5 Time Until Treatment for Cystic Fibrosis

In order to look at overall program efficiency with respect to treating newborns identified with cystic fibrosis, programs were asked to report the number of days from birth until treatment. The general definition of treatment used in the questionnaire was, “positive sweat chloride test” and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 10.05.

10.6 Historic Data

In order to document the value of screening for CF, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of cases detected over the years. These data are given in Table 10.06. Since the programs involved are young, the data are considered to be accurate and comprehensive but the validity may suffer from the same problems as noted in other chapters.

10.7 Total Newborns Screened

Table 10.07 gives a summation of the data contained in Tables 10.02 and 10.03 along with the number of births in each state or territory as reported in Table 1.01. This table was included so that a tabulation of these data could be viewed without having to refer to several tables.

10.8 Summation of Results

Table 10.08 sums the significant testing data from this Chapter. It includes the total births from Table 1.01, the total infants reported as being screened, and the testing data reported in Tables 10.02 and 10.03. Comparative percentages have been provided at the request of a number of readers, but in at least one case these data appear to be based on estimates or on data that contain duplicate information causing the total compliance percentage to exceed 100%. Hopefully, future data reports will be able to correct these data.

Table 10.01: Cystic Fibrosis - Laboratory Testing

State/Territory	Testing Method	Secondary Testing Method	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Sweat Chloride Testing</i>
1 Colorado	FIA (DELFIA)		≥ 90 ng/mL IRT on initial specimen	≥ 70 ng/mL IRT on recall specimens
2 Connecticut	Immunoreactive	PCR ^a F508 mutation	≥ 90 ng/mL	NA
3 Massachusetts	IRT	Assay for 27 mutations	none	none
4 Montana	Fluorometric	Sweat chloride/DNA	1st specimen ≥ 100 ng/mL	Second test @ 3-4 wks of age > 80ng/mL.
5 Wisconsin	DELFIA	PCR - ΔF508	a	a
6 Wyoming	DELFIA		≥ 90 ng/mL IRT initial	≥ 70 ng/mL IRT on recall specimens
7 NeoGen Screening	ICN	IRT > 90 ng/mL perform ΔF508 analysis.	IRT > 130 ng/mL and no copies of ΔF508	For 2 copies of ΔF508 (homozygous), a sweat test is recommended for confirmation. For 1 copy fo ΔF508 (heterozygous), a sweat test + additional DNA analysis is recommended . When IRT > 90 ng/mL, a sweat test on second spec., a sweat test + additional DNA analysis is recommended.

a = sweat test to be performed at one month of age if one or two ΔF508 allele(s) are reported on newborn screen report.

Table 10.02: Initial Screening Results - Cystic Fibrosis

State/Territory	Number of Newborns Screened	Number of Newborns with <u>NOT NORMAL</u> Tryspin Results	Number of Newborns also having <u>NOT NORMAL</u> DF508	Number of Newborns with final <u>NOT NORMAL</u> Classification Lost to follow-up	Number of Newborns Confirmed with Cystic Fibrosis
1 Colorado	63,219	670	DA	10 a	9
2 Connecticut	20,841	287	NA	NA	NA
3 Massachusetts	81,601	3,077	252	8 f	31
4 Montana	5,197	16	0	5	1
5 Wisconsin	66,614	26	144	5	16 b
6 Wyoming	5,480	27	NC	3	2
7 NeoGen Screening	140,789	258	20	38 c	20
TOTAL	d	d	d	d	79 e

a = certified letters to mothers, no further information; **b** = one infant not identified by NBS (2 non F508 mutations);

c = physician notification or baby ctb; **d** = totals not given - too many states reported inaccurate or inappropriate data;

e = total given does not include all states; **f** = 5 died, 2 moved out of country, 1 multiple no-show at pcip.

Table 10.03: Second Screens for Cystic Fibrosis

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen R	Newborns Receiving Discretionary Second Screens D	Newborns Receiving Repeat Second Screens P	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Trypsin Results				Number of Newborns with <u>NOT NORMAL</u> DF508				Number of Newborns with <u>NOT NORMAL</u> Classification Lost to follow-up				Number of Newborns Confirmed with Cystic Fibrosis			
					R	D	P	Total	R	D	P	Total	R	D	P	Total	R	D	P	Total
1 Colorado	60,530		N/C	60,530				0	DA		DA		NC		NC		0		NA	0
2 Connecticut			287	287			56	56			4	4			0	0			2	2
3 Massachusetts																				
4 Montana				0																
5 Wisconsin		1,196	1,103	2,299		0	0	0												
6 Wyoming	DA		NC		DA		NC		DA		NC		DA		NC		DA		NC	
7 NeoGen Screening		1,984	1,546	3,530 a	DA		NC		DA		NC		DA		NC		DA		NC	
Total	b	60,530	3,180	2,936	c			c				c				c	0	0	2	2 d

a = these numbers are overall; **b** = totals given reflect program responses and should be viewed as estimates only, given the number not reporting and the number of caveats listed;

c = totals not given - too many programs reported inaccurate or inappropriate data; **d** = total given does not include all states.

Table 10.04: Cases of Cystic Fibrosis

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Colorado											4	5	4	5	9		
2 Connecticut	1	1											1	1	2			
3 Massachusetts															31	¹		
4 Montana															1	¹		
5 Wisconsin	5	9					1				1		7	9	16			
6 Wyoming		2											0	2	2			
7 NeoGen Screening	10	8										2	10	10	20			
TOTAL	16	20	0	0	0	0	1	0	0	0	5	7	22	27	81	0	0	0

¹did not report a breakdown of Race/Ethnicity.

²total reported is from Table 10.02 and Table 10.03; therefore total does not equal or may differ from data given in Table 10.04 and denotes a problem in reliability.

³Ethnicity counted as a Race and is included in total.

Table 10.05: Days from Birth Until Treatment Initiated for Cystic Fibrosis

Total detected on initial and second screens

(blank spaces mean that no infants fell into these categories)

State/Territory	≤ 3	4	5	6	7	8	9	10	11	12	13	14	15-21	>21	Unk.	TOTAL
1 Colorado ^a													3	4	2	9
2 Connecticut														2		2
3 Massachusetts															31	31
4 Montana															1	1
5 Wisconsin								1	2				2	6	5	16
6 Wyoming															2	2
7 NeoGen Screening															20	20

¹did not report a breakdown of Days from Birth.

a = Co. does not refer for diagnostic testing until two screens have been abnormal for CF.

Table 10.06: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Cystic Fibrosis		
	Year Started	Total Years	Total Cases
1 Colorado	Jul-1987	12 1/2	188
2 Connecticut	May-1993	7 1/2	44
3 Massachusetts	1999	1	~60
4 Montana	1992	8	15
5 Wisconsin	1994	6	100
6 Wyoming	Sept-1998	2 1/4	22
7 NeoGen Screening NR			

Table 10.07: Total Newborns Screened for Cystic Fibrosis

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Trypsin Results	Number of Newborns also having <u>NOT NORMAL</u> DF508	Number of Newborns with final <u>NOT NORMAL</u> Classification Lost to follow-up	Number of Newborns Confirmed with Cystic Fibrosis
1 Colorado	65,679	670	DA	10	9
2 Connecticut	43,370	343	4	0	2
3 Massachusetts	82,673	3,077	252	8	31
4 Montana	10,927	16	0	5	1
5 Wisconsin	68,250	26	144	5	16
6 Wyoming	5,847	27	DA	3	2
7 NeoGen Screening	144,319	258	20	38	20
TOTAL	421,065	4,417	440	69	81

a = total given does not include all states.

Table 10.08: Summation Results of Testing for Cystic Fibrosis

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screen calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary 2nd Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Colorado	65,679	63,219	96.25%	60,530	92.16%				
2 Connecticut	43,370	20,841	48.05%					287	0.66%
3 Massachusetts	82,673	81,601	98.70%						
4 Montana	10,927	5,197	47.56%						
5 Wisconsin	68,250	66,614	97.60%			1,196	1.75%	1,103	1.62%
6 Wyoming	5,847	5,480	93.72%						
7 NeoGen Screening	144,319	140,789	97.55%			1,984	1.37%	1,546	1.07%
TOTAL	a	383,741	b		b	3,180	b	2,936	b

* Percentage > 100% denotes inability of program to separate Newborns screened from specimens received; **a** = totals should be viewed as a rough estimate due to inability of many states, to know actual number of newborns screened. Many programs count samples received but cannot eliminate duplication when multiple samples are received; **b** = cannot be calculated due to inaccurate data.

11. Tyrosinemia

11.0 Introduction

Screening for tyrosinemia is now formally included in only a few screening programs. Some programs, however, check tyrosine levels when confirming hyperphenylalaninemia on newborns with elevated phenylalanine levels detected through newborn screening. All data reported here were collected from questionnaires submitted to programs as noted in Chapter 3. Only programs with tyrosinemia screening formally included in their testing schemes are listed in the Tables.

11.1 Laboratory Procedures

In order to identify laboratory procedures in use, programs were asked to report their laboratory protocols and definitions for samples considered 'not normal.' These data are reported in Table 11.01.

11.2 Initial Screening Results

In order to ascertain the effectiveness of screening for tyrosinemia, programs were asked to report their findings on initial screening. Initial screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of infants screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated. Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 11.02. Programs wishing to explain some of their responses included information as footnotes to the table. Because some requested data were not submitted, some care should be exercised in their use.

11.3 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are reported in Table 11.03

and are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when a second test is performed without regard for the initial screening result. Programs that requested a second screen because of either a certain result on the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as 'repeat screens' and these are also included.

For each case, the data requested included the total infants tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding. It should be recognized that this is a different way of reporting these data from previous Reports and may, therefore, be somewhat confusing. It should be recognized that this is a different way of reporting these data than previously and may, therefore, be somewhat confusing. It is hoped that these data better represent the findings from additional screens. Once again, the reader is cautioned about using the data without regard to notes of explanation.

11.4 Cases of Tyrosinemia Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of tyrosinemia, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 11.04. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

11.5 Time Until Treatment for Tyrosinemia

In order to look at overall program efficiency with respect to treating newborns identified with tyrosinemia, programs were asked to report the number of days from birth until treatment. The definition of treatment used in the questionnaire was “initiation of dietary or drug regimen” and programs using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 11.05

11.6 Historic Data

In order to document the value of screening for tyrosinemia, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of cases detected over the years. These data are given in Table 11.06.

11.7 Total Newborns Screened

Table 11.07 gives a summation of the data contained in Tables 11.02 and 11.03 along with the number of births in each state or territory as reported in Table 1.01. This table was included at the request of a number of screeners who wished to see a tabulation of these data without having to refer to several tables.

11.8 Summation of Results

Table 11.08 sums the significant testing data from this Chapter. It includes the total births from Table 1.01, the total infants reported as being screened, and the testing data reported in Tables 11.02 and 11.03. Comparative percentages have been provided at the request of a number of readers, but in several cases, these data appear to be based on estimates or on data that contain duplicate information causing the total compliance percentage to exceed 100%.

Table 11.01: Tyrosinemia - Laboratory Testing

State/Territory	Testing Method	Other	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Immediate Serum Follow-up</i>
1 Georgia	BIA		Tyrosine >12 mg/dL but < 20 mg/dL	Tyrosine > 20 mg/dL on initial test.
2 Maryland	BIA Assay	HPLC	10-12 mg/dL	> 12 mg/dL
3 Massachusetts		MS/MS		
4 North Carolina		MS/MS	Tyrosine \geq 300 μ M	Tyrosine \geq 900 μ M

Table 11.02: Initial Screening Results -Tyrosinemia

State/Territory	Number of Newborns Screened	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Result Lost to follow-up	Number of Newborns Confirmed with Tyrosinemia
1 Georgia	189,498			
2 Maryland	1,871	1		
3 Massachusetts	81,372	76		
4 North Carolina	120,750			
TOTAL	393,491	77	0	0

Table 11.03: Second Screens for Tyrosinemia

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed with Tyrosinemia			
	R	D	P		R	D	P	Total	R	D	P	Total	R	D	P	Total
1 Georgia																
2 Maryland	61,152	0		61,152	2			2					2			2
3 Massachusetts							57	57								
4 North Carolina																
Total	a 61,152	0	0	61,152 b	2		57	59 b	0	0	0	0 b	2		0	2 b

a = totals given reflect program responses and should be viewed as estimates only given, the number not reporting and the number of caveats listed; **b** = total given does not include all states.

Table 11.04: Cases of Tyrosinemia

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic			
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Total	Male	Female	Total
	1 Georgia																	
2 Maryland	2												2	0	2			
3 Massachusetts																		
4 North Carolina																		
TOTAL	2												2	0	2			

Table 11.06: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Tyrosinemia		
	Year Started	Total Years	Total Cases
1 Georgia	September-78	21 1/4	6
2 Maryland	1977	23	4
3 Massachusetts NR			
4 North Carolina NR			

Table 11.07: Total Newborns Screened for Tyrosinemia

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Result Lost to follow-up	Number Newborns Confirmed with Tyrosinemia
1 Georgia	133,524	0	0	0
2 Maryland	69,574	3		2
3 Massachussetts	82,673	133		0
4 North Carolina	121,347	0	0	0
TOTAL	407,118	136	0	2 a

a = babies were twins with Tyrosinemia I, both identified clinically as no repeat specimens received.

Table 11.08: Summation Results of Testing for Tyrosinemia

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screen calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary 2nd Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Georgia	133,524	189,498	* 141.92%						
2 Maryland	69,574	1,871	2.69%	61,152	87.89%				
3 Massachusetts	82,673	81,372	98.43%						
4 North Carolina	121,347	120,750	99.51%						
TOTAL a	407,118	393,491	b	61,152	b				

* Percentage > 100% denotes inability of program to separate Newborns screened from specimens received; **a** = totals should be viewed as a rough estimate due to inability of many states, to know actual number of Newborns screened. Many programs count samples received but cannot eliminate duplication when multiple samples are received; **b** = cannot be calculated due to inaccurate data.

12. Toxoplasmosis

12.0 Introduction

Screening for toxoplasmosis is included in only two screening programs. Its inclusion is somewhat different from the other disorders included in screening programs since it detects an infectious disease. The goal of newborn screening is early identification and treatment to minimize the risk of blindness, mental retardation, or other related problems. Both states involved in screening receive laboratory services from the same laboratory in Massachusetts and so there are no differences in screening protocols or definitions. The data reported here was collected from questionnaires submitted to programs as noted in Chapter 3. Only programs with tyrosinemia screening formally included in their testing schemes are listed in the Tables.

12.1 Laboratory Procedures

In order to identify laboratory procedures in use, programs were asked to report their laboratory protocols and definitions for samples considered 'not normal.' These data are reported in Table 12.01.

12.2 Initial Screening Results

In order to ascertain the effectiveness of screening for toxoplasmosis, programs were asked to report their findings on initial screening. Initial screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of newborns screened and confirmed, versus the number of positive tests reported, the efficiency of the testing protocol may be evaluated. Additionally, programs were asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported in Table 12.02. If programs wished to explain some of their responses the information is included as a footnote to the table.

12.3 Second Screen Results

Please see Section 3.6 for a more complete discussion of the data on second screening tests. Second screen data are reported in Table 12.03 and are divided on the basis of whether the second screens are required on all newborns ('required') or are obtained on some other basis ('discretionary') when a second test is performed without regard for the initial screening result. Programs that requested a second screen because of either a certain result on the first screen that acted as impetus for a second screen or because of an unsatisfactory first screen (usually due to sample condition or early collection) were asked to report these data as 'repeat screens' and these are also included. For each case, the data requested included the total newborns tested, the total with findings considered not normal, and the number of confirmed cases divided by type of clinical finding.

12.4 Cases of Toxoplasmosis Divided by Sex and Race/Ethnicity

Programs were asked to report the total of all confirmed cases of toxoplasmosis, whether detected on first or second screen, divided by sex and race/ethnicity. The term race/ethnicity was used because some states with high Hispanic populations maintain Hispanic data in much the same way that other states maintain racial data. These data are reported in Table 12.04. For comparative data about the breakdown of births in all states and territories, the reader is referred to the tables in Chapter 1.

12.5 Time Until Treatment for Toxoplasmosis

In order to look at overall program efficiency with respect to treating newborns identified with toxoplasmosis, programs were asked to report the number of days from birth until treatment. The definition of treatment used in the questionnaire was "initiation of dietary or drug regimen" and programs

using a different definition were asked to report their definition for inclusion as a footnote to the tabulation. Similarly programs were asked to provide footnote information for any cases not treated within the first 21 days of life. These data are reported in Table 12.05.

12.6 Historic Data

In order to document the value of screening for toxoplasmosis, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of cases detected over the years. These data are given in Table 12.06.

12.7 Total Newborns Screened

Table 12.07 gives a summation of the data contained in Tables 12.02 and 12.03 along with the number of births in each state or territory as reported in Table 1.01. This table was included for those who wish to see a tabulation of these data without having to refer to several tables.

12.8 Summation of Results

Table 12.08 sums the significant testing data from this Chapter. It includes the total births from Table 1.01, the total newborns reported as being screened, and the testing data reported in Tables 12.02 and 12.03. Comparative percentages have been provided to give an idea of program coverage.

Table 12.01: Toxoplasmosis - Laboratory Testing

State/Territory	Testing Method	Secondary Test List Method	Definition of NOT NORMAL (mg/dL) <i>Requiring Further Filter Paper Testing</i>	Definition of NOT NORMAL (mg/dL) <i>Requiring Immediate Serum Follow-up</i>
1 Massachusetts	IgM	IgG	≥ 0.100 OD	≥ 0.100 OD
2 New Hampshire	IgM	IgG	≥ 0.100 OD	> 0.100 OD

Table 12.02: Initial Screening Results - Toxoplasmosis

State/Territory	Number of Newborns Screened	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Result Lost to follow-up	Number of Newborns Confirmed with Toxoplasmosis
1 Massachusetts	82,703	22	0 N/A	1
2 New Hampshire	13,879	1	0	0
TOTAL	96,582	23	0	1 a

a = total given does not include all states.

Table 12.03: Second Screens for Toxoplasmosis

R = Required Second Screens D = Discretionary Second Screens P = Repeat Second Screens

State/Territory	Newborns Receiving Required Second Screen	Newborns Receiving Discretionary Second Screens	Newborns Receiving Repeat Second Screens	Total 2nd Screens	Number of Newborns with <u>NOT NORMAL</u> Test Results				Number of Newborns with <u>NOT NORMAL</u> Test Results Lost to Follow-up				Number of Newborns Confirmed with Toxoplasmosis			
	R	D	P		R	D	P	Total	R	D	P	Total	R	D	P	Total
1 Massachusetts			8,818	8,818			5	5				0				0
2 New Hampshire		815		815												
Total		815	8,818	9,633			5	5				0				0

Table 12.04: Cases of Toxoplasmosis

Total detected on initial and second screens

Subdivided by Sex and Race/Ethnicity

State/Territory	White		Black		Asian or P. Islander		Native American		Other		Unknown		Total.....		Hispanic		Total	
	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female		
	Total																	
1 Massachusetts												1	0	1	1			
2 New Hampshire																		
TOTAL															1			

Table 12.06: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	Toxoplasmosis		
	Year Started	Total Years	Total Cases
1 Massachusetts	1986	14	120
2 New Hampshire	1988	12	10

Table 12.07: Total Newborns Screened for Toxoplasmosis

State/Territory	Total Births	Number of Newborns with <u>NOT NORMAL</u> Test Results	Number of Newborns with <u>NOT NORMAL</u> Test Result Lost to follow-up	Number of Newborns Confirmed with Toxoplasmosis
1 Massachusetts	82,673	27	0	1
2 New Hampshire	13,987	1	0	0
TOTAL	96,660	28	0	1

Table 12.08: Summation Results of Testing for Toxoplasmosis

State/Territory	Total Births	Total Newborns Receiving Initial Screen	% of Newborns Receiving Initial Screen calculated	Total Newborns Receiving Required 2nd Screen	% of Newborns Receiving Required 2nd scrn. (calc'd)	Total Newborns Receiving Discretionary Screen	% of Newborns Receiving Discretionary 2nd scrn. (calc'd)	Total Newborns Receiving Repeat 2nd Screen	% of Newborns Receiving Repeat 2nd scrn. (calc'd)
1 Massachusetts	82,673	82,703	* 100.04%					8,818	10.67%
2 New Hampshire	13,987	13,879	99.23%			815	5.83%		
TOTAL	96,660	96,582	116.82%			815	5.83%	8,818	10.67%

* Percentage > 100% denotes inability of program to separate Newborns screened from specimens received.

13. Hemoglobinopathies

13.0 Introduction

Screening for hemoglobinopathies, particularly those involving sickle hemoglobin, have been included in U.S. screening programs since the 1970's, but the majority of screening programs added this testing in the late 1980's. Initially the screening technique included hemoglobin electrophoresis in basic and alkaline medium to identify potentially affected newborns. Confirmation testing was recommended at 2 - 3 months when fetal hemoglobin concentrations decreased to a point where it no longer interfered with the diagnostic tests. As isoelectric focusing techniques improved, this technique became popular because of its relatively low cost and relative ease of interpretation. Some programs have also included high performance liquid chromatography (HPLC) as either a primary or ancillary screening technique, and some have included DNA as a confirmatory procedure. All data reported here were collected from questionnaires submitted to programs as noted in Chapter 3. Only programs with hemo-globinopathy screening included in their testing scheme are listed in the Tables.

13.1 Laboratory Procedures

In order to identify laboratory procedures in use, programs were asked to report their laboratory protocols for both the primary method and any ancillary testing. These data are reported in Table 13.01.

13.2 Initial Screening Results

In order to ascertain the effectiveness of screening for hemoglobinopathies, programs were asked to report their findings on initial screening for a selected group of hemoglobinopathy findings. Initial screening was defined as the first reportable test, and programs were asked not to include duplicate screenings. By reviewing the number of newborns screened and confirmed, and the number lost to follow-up,

the efficiency of the testing protocol may be evaluated. Programs were also asked to report the number of positive patients 'lost to follow-up' as a means of evaluating the follow-up procedures. These data are reported divided by Race in Table 13.02. Programs wishing to explain some of their responses included information as footnotes to the table. Care should be taken in interpreting these data, since not all programs are designed to confirm and follow all reported results. For example, most programs do not confirm results considered normal such as Hb F,A.

Information was also requested on second testing and repeat testing as with other disorders, but the responses were limited to a handful of programs and it was apparent that the reported data were questionable. For this reason, no data were tabulated on second testing and repeat testing. Comments from readers on the usefulness of this type of information would be helpful for future Newborn Screening Data Reports.

13.3 Time Until Diagnosis for FS Cases (Sickle Cell Anemia)

In order to look at overall program efficiency with respect to speed of diagnosis, programs were asked to report the number of days from birth until diagnosis. For purposes of this Report, diagnosis was defined as, "confirmation of the screening results (usually outside of the original screening environment - e.g. confirmation of sickle cell anemia by repeat electrophoretic techniques at 2-3 months; DNA testing at 1 week; etc.)." Programs using a different definition were asked to give the definition and it is included as a footnote to the Table. These data are reported in Table 13.03.

13.4 Time Until Treatment for FS Cases (Sickle Cell Anemia)

So that programs may know the policies and actions of other programs with respect to treatment for sickle cell anemia, data were gathered relative to time until treatment. Treatment was defined as, "initiation of ... drug regimen." These data are reported in Table 13.04. Similar data were not requested for other sickle cell diseases since this disease was considered sufficiently representative of program actions.

13.5 Time to Diagnosis for FSA Cases (S- β ⁺-thalassemia)

Programs were asked to report the time to diagnosis for FSA cases since these cases may face a health threat and laboratory results may sometimes be confused with FS and FAS at the time of initial screening. These data are reported in Table 13.05.

13.6 Time to Diagnosis for FSC Cases (SC-Disease)

Programs were asked to report the time to diagnosis for FSC cases and these data are reported in Table 13.06.

13.7 Time to Diagnosis for FSE Cases (SE-Disease)

Programs were asked to report the time to diagnosis for FSE cases and these data are reported in Table 13.07.

13.8 Time to Diagnosis for FSD Cases (SD-Disease)

Programs were asked to report the time to diagnosis for FSD cases and these data are reported in Table 13.08.

13.9 Time to Diagnosis for FAS Cases (Sickle Cell Trait)

Programs were asked to report the time to diagnosis for FAS cases and these data are reported in Table 13.09.

13.10 Program Follow-up

In order to look at the activities of programs related to follow-up of sickle trait, programs were asked to report related information. Specifically, programs were asked to indicate whether or not they maintained an aggressive follow-up program for sickle trait, whether there was extra staff available for this follow-up, and any other comments appropriate to follow-up of sickle trait. These responses are tabulated in Table 13.10.

13.11 Historic Data

In order to document the value of screening for hemoglobinopathies, or more specifically for sickle hemoglobinopathies, programs were asked to supply whatever historic data they could on their screening program. In particular, they were asked to give the date the program began and the number of cases detected for each different type of case over the years. These data are given in Table 13.11.

Table 13.01: Hemoglobinopathy Testing Methodology

State/Territory	Primary Laboratory Method	Secondary Laboratory Method
1 Alabama	Electrophoresis	HPLC
2 Arizona	IEF	HPLC, Isoscan run as secondary method
3 Arkansas	IEF	Citrate agar electrophoresis
4 California	HPLC	
5 Colorado	IEF	IEF & HPLC to confirm
6 Connecticut	HPLC	IEF
7 Delaware	IEF	DA
8 District of Columbia	IEF	DNA Analysis
9 Florida	IEF	HPLC
10 Georgia	IEF	HPLC, Citrate Agar
11 Hawaii	IEF	HPLC
12 Illinois	HPLC	
13 Indiana	IEF	Citrate Agar, Beckman/Paragon, DNA
14 Iowa	IEF	HPLC
15 Kansas	IEF	
16 Kentucky	IEF	N/A
17 Louisiana	HPLC	IEF
18 Maine	IEF	Citrate Agar
19 Maryland	IEF	HPLC
20 Massachusetts	IEF	Citrate Agar Electrophoresis
21 Michigan	HPLC	IEF
22 Minnesota	IEF	
23 Mississippi	IEF	IEF
24 Missouri	IEF	HPLC
25 Montana	IEF	HPLC done by NM Newborn Screening Lab
26 Nebraska	HPLC	
27 Nevada	Electrophoresis	
28 New Hampshire	IEF	Citrate Agar Electrophoresis
29 New Jersey	IEF	HPLC
30 New Mexico	HPLC column chromatography	IEF
31 New York	Cellulose Acetate Electrophoresis	Citrate Agar Electrophoresis, IEF
32 North Carolina	IEF	HPLC
33 North Dakota	IEF	HPLC
34 Ohio	IEF	
35 Oklahoma	IEF	Whole Blood = IEF, Solubility = Citrate Agar
36 Oregon	IEF	HPLC
37 Pennsylvania	IEF	
38 Rhode Island	IEF	Citrate Agar Electrophoresis
39 South Carolina	IEF	HPLC
40 Tennessee	IEF	
41 Texas	IEF	IEF, HPLC, DNA confirmation on spots.
42 Vermont	IEF	Citrate Agar Electrophoresis
43 Virginia	IEF	HPLC
44 Washington	IEF	HPLC & PCR/DNA analysis
45 West Virginia	IEF	
46 Wisconsin	IEF	HPLC
47 Wyoming	IEF	IEF & Citrate Agar
48 Puerto Rico	Cellulose Acetate at pH 8.4	Citrate Agar electrophoresis at pH 6.2
49 Virgin Islands	IEF	HPLC

Table 13.02: INITIAL HEMOGLOBIN RESULTS (continued)

S = Suspected

C = Confirmed

State/Territory	Number of Newborns Screened	FA + Other																	
		White		Black		Asian or P. Islander		Native Am.		Other		Unknown		Total Reported		Hispanic		Lost to Follow-up	
		S	C	S	C	S	C	S	C	S	C	S	C	S	C	S	C		
1 Alabama	61,877																		
2 Arizona	81,956										132	79	132	79					
3 Arkansas	36,703	29		135		12				2		3		183 ¹			2		DA
4 California	527,297	117		61		66		2		27		11		576 ¹	N/A		292		N/A
5 Colorado	63,219											80	80	80	80				
6 Connecticut	43,734																		
7 Delaware	11,714	5	5	4	4					1	1			10	10		1	1	
8 District of Columbia	15,125											1		1	0				
9 Florida	204,030																		
10 Georgia	191,145	4																	
11 Hawaii	17,612	8		18		305		1		59				391 ^a			23		N/A
12 Illinois	188,164													0					
13 Indiana	87,639	164		361		26				43				615 ¹			21		
14 Iowa	38,141											131	83	131	83				N/C
15 Kansas	39,031											112	15	112	15				97
16 Kentucky	54,515																		
17 Louisiana	67,843													0	0				DA
18 Maine	3,194											8		8					
19 Maryland	72,390	81		56		37				24		27		232 ¹			7		
20 Massachusetts	82,703																		
21 Michigan	134,022													0					
22 Minnesota	68,402													0					
23 Mississippi	44,075													0	0				
24 Missouri	78,658	84		29		2				2		4		124 ¹			3		

Continued

Table 13.02: INITIAL HEMOGLOBIN RESULTS (continued)

S = Suspected C = Confirmed

State/Territory	Number of Newborns Screened	FA + Other														Total Reported	Hispanic		Lost to Follow-up	
		White		Black		Asian		Native Am.		Other		Unknown		S	C		S	C		
		S	C	S	C	S	C	S	C	S	C	S	C							
25 Montana	10,917											20	20	20	b	20			20	
26 Nebraska	24,863											39	23	39		23			16	
27 Nevada	30,659	26	26	4	4	2	2			27	27			77	¹	77	¹	18	18	
28 New Hampshire	5,018											17	17	17		17				
29 New Jersey	112,241											995		995		DA				
30 New Mexico	25,865	15		6		5	2		2		4			51	¹			17		
31 New York	258,449	233		865		179			110		N/A			1,540	¹			153	N/C	
32 North Carolina	120,750	117		77		9	9		29		6			247						
33 North Dakota	1																			
34 Ohio	160,566											233	50	233		50				
35 Oklahoma	52,760											77	35	77		35			31	
36 Oregon	46,879	53		4		6	1					22		86			24			
37 Pennsylvania												214		214						
38 Rhode Island	13,150											29		29	a					
39 South Carolina	52,176											163		163		DA				
40 Tennessee	79,539													0						
41 Texas	355,100	225		92		20			43		29			840	¹			431		
42 Vermont	6,040		10											0		10				
43 Virginia	99,410	183		649		65	1		75		25			998				25		
44 Washington	76,886	189	102	157	9	182	9	4	3	271	33			803	a	156	a	73	36	
45 West Virginia	10,818	10		10						2				22						
46 Wisconsin	66,614	2		1										3						
47 Wyoming	5,480											8	7	8		7				
48 Puerto Rico	57,048									11	11			11		11		11	11	4
49 Virgin Islands	1,851											2	2	2		2				
Total	3,850,489	1,545	143	2,529	17	916	11	20	3	728	72	2,392	411	9,074	^S	675	^C	1,101	66	168

¹ Ethnicity counted as a Race and is included in total.
a = Hb Bart's data; **b** = 2 FAX Barts, 6 FAX probable G variants, 12 FAX.

Table 13.03: Summation of Confirmed FS Cases (Sickle Cell Anemia) by Days from Birth Until Diagnosis

State/Territory	0-15	16-30	31-45	46-60	61-75	76-90	> 90	Unknown	Total
1 Alabama	15	9	5	3	1		3	2	38
2 Arizona			1	1			1		3
3 Arkansas	5	2	2	1		1	1		12
4 California	1	24	6	1	1			15	48
5 Colorado								6	6
6 Connecticut	10		1						11
7 Delaware		5							5
8 District of Columbia									
9 Florida	4	14	15	6	10	7	14		70
10 Georgia								105	105
11 Hawaii							1		1 ^a
12 Illinois	1	3	7	11	8	4	11	17	62
13 Indiana								21	21
14 Iowa	1	1							2
15 Kansas								11	11
16 Kentucky ^{NR}									
17 Louisiana	49	11	2				2		64
18 Maine									
19 Maryland		4	3	6	3	1	3	1	21
20 Massachusetts									
21 Michigan	3	3	4		2	4	10	1	27
22 Minnesota		2	4	1					7
23 Mississippi		18	6				2	1	27
24 Missouri		4	4	1	1	1	10	3	24
25 Montana									NC
26 Nebraska	1	1							2
27 Nevada		4							4
28 New Hampshire									
29 New Jersey	3	20	12	8	3	2	2	1	51
30 New Mexico									
31 New York	34	42	22	8	14	4	19	2	145
32 North Carolina									ND
33 North Dakota									
34 Ohio								34	34
35 Oklahoma		3		2	1				6
36 Oregon									
37 Pennsylvania	10	7	4	3	2	1	6	3	36
38 Rhode Island									
39 South Carolina	2	11	8	3	2	1	3	4	34
40 Tennessee									
41 Texas									NC
42 Vermont									
43 Virginia	17	4							21
44 Washington		4							4
45 West Virginia	2								2
46 Wisconsin		4	3		1				8
47 Wyoming									
48 Puerto Rico	1	2							3
49 Virgin Islands	7								7

^a = infant was Hb-S with hereditary persistent fetal hgb.

**Table 13.04: Summation of Confirmed FS Cases (Sickle Cell Anemia) by Days from Birth
Until Treatment Initiated**

State/Territory	0-15	16-30	31-45	46-60	61-75	76-90	> 90	Unknown	Total
1 Alabama	15	11	4	4	1		1	2	38
2 Arizona			1		1		1		3
3 Arkansas	1	2	2	1	2	1	3		12
4 California	4	8	10	13	5	1	1	6	48
5 Colorado								6	6
6 Connecticut	10		1						11
7 Delaware		2						3	5
8 District of Columbia									
9 Florida									
10 Georgia								105	105
11 Hawaii								1	1 a
12 Illinois	1	3	7	11	8	4	11	17	62
13 Indiana								21	21
14 Iowa							2		2
15 Kansas								11	11
16 Kentucky NR									
17 Louisiana	47	9	3						59
18 Maine									
19 Maryland		4		3			1	55	63
20 Massachusetts									
21 Michigan	2	4	1	3	1	1	12	3	27
22 Minnesota	1	5	1						7
23 Mississippi		18	6				2	1	27
24 Missouri	2	2	2	3	3	1	5	6	24
25 Montana									NC
26 Nebraska		1	1						2
27 Nevada		4							4
28 New Hampshire									
29 New Jersey		15	5	14	5			12	51
30 New Mexico									
31 New York	135	7	2				1		145
32 North Carolina									N/D
33 North Dakota									
34 Ohio								34	34
35 Oklahoma	1	3		1	1				6
36 Oregon									0
37 Pennsylvania	4	7	10	5	3	2	2	3	36
38 Rhode Island									
39 South Carolina		3		2	1			28	34
40 Tennessee									
41 Texas									NC
42 Vermont									
43 Virginia				20		1			21
44 Washington		2	1		1				4
45 West Virginia	2								2
46 Wisconsin		1	1	3				3	8
47 Wyoming									
48 Puerto Rico			1	2					3
49 Virgin Islands									

a = Hb-S & hereditary fetal hgb, infant not placed on medications, is being monitored by hematologist.

Table 13.05: Summation of Confirmed FSA Cases (S-b⁺-thalassemia) by Days from Birth Until Diagnosis

State/Territory	0-15	16-30	31-45	46-60	61-75	76-90	> 90	Unknown	Total
1 Alabama	1			1					2
2 Arizona		1							1
3 Arkansas						1	2		3
4 California		1	1		5	1	2		10
5 Colorado									
6 Connecticut	1	2							3
7 Delaware									NC
8 District of Columbia									
9 Florida		4	1	3	1	4	3		16
10 Georgia									
11 Hawaii									
12 Illinois		1	1		2	1	4	5	14
13 Indiana								3	3
14 Iowa	1		2				1		4
15 Kansas									
16 Kentucky NR									
17 Louisiana	5	1							6
18 Maine									
19 Maryland			1					1	2
20 Massachusetts									
21 Michigan		2	1	1	1	1	4		10
22 Minnesota									
23 Mississippi		1	1	1					3
24 Missouri									
25 Montana									NC
26 Nebraska									
27 Nevada									
28 New Hampshire									NC
29 New Jersey		3							3
30 New Mexico				2					2
31 New York		5	1				1		7
32 North Carolina									ND
33 North Dakota									
34 Ohio									NC
35 Oklahoma									
36 Oregon									
37 Pennsylvania					1			1	2
38 Rhode Island									
39 South Carolina	1	3	1	2			1		8
40 Tennessee									NR
41 Texas									NC
42 Vermont									
43 Virginia	2	2		1					5
44 Washington			1				1		2
45 West Virginia									ND
46 Wisconsin		1							1
47 Wyoming									
48 Puerto Rico									
49 Virgin Islands									

Table 13.06: Summation of Confirmed FSC Cases (SC-Disease) by Days from Birth Until Diagnosis

State/Territory	0-15	16-30	31-45	46-60	61-75	76-90	> 90	Unknown	Total
1 Alabama	8	8	1	1	1		2	1	22
2 Arizona			1				1		2
3 Arkansas	8	3	3			1	2		17
4 California	2	2	4	8	2	2	3	5	28
5 Colorado			4						4
6 Connecticut	8	3							11
7 Delaware	2			1					3
8 District of Columbia									
9 Florida	3	8	12	8	5	3	20	5	64
10 Georgia									
11 Hawaii		1							1
12 Illinois	1	2	5	7	2	1	7	4	29
13 Indiana								3	3
14 Iowa									
15 Kansas								2	2
16 Kentucky NR									
17 Louisiana	15	4							19
18 Maine									
19 Maryland		1	1		1	1	1	1	6
20 Massachusetts									
21 Michigan				2	5	1	6		14
22 Minnesota		2	1	1				1	5
23 Mississippi		7	1				7		15
24 Missouri		3	2	3	1	1	5		15
25 Montana									NC
26 Nebraska		1							1
27 Nevada		3							3
28 New Hampshire									
29 New Jersey	4	10	11	3		1			29
30 New Mexico									
31 New York	16	28	13	6	4	1	9	3	80
32 North Carolina									ND
33 North Dakota									
34 Ohio									NC
35 Oklahoma					1				1
36 Oregon								1	1
37 Pennsylvania	1	2	5	2	3			4	17
38 Rhode Island									
39 South Carolina	3	2	4		1		2	3	15
40 Tennessee									
41 Texas									NC
42 Vermont									
43 Virginia	6	5	3				2		16
44 Washington	1		1						2
45 West Virginia									ND
46 Wisconsin		1	1	1					3
47 Wyoming									
48 Puerto Rico	2	4							6
49 Virgin Islands	6								6

Table 13.07: Summation of Confirmed FSE Cases (SE-Disease) by Days from Birth Until Diagnosis

State/Territory	0-15	16-30	31-45	46-60	61-75	76-90	> 90	Unknown	Total
1 Alabama									
2 Arizona									
3 Arkansas									
4 California			2					1	3
5 Colorado									
6 Connecticut									
7 Delaware									NC
8 District of Columbia									
9 Florida									
10 Georgia									
11 Hawaii									
12 Illinois									
13 Indiana									
14 Iowa									
15 Kansas								1	1
16 Kentucky NR									
17 Louisiana									ND
18 Maine									
19 Maryland									
20 Massachusetts									
21 Michigan									
22 Minnesota									
23 Mississippi									
24 Missouri									
25 Montana									NC
26 Nebraska									
27 Nevada									
28 New Hampshire									
29 New Jersey			1						1
30 New Mexico									
31 New York	1	1							2
32 North Carolina									ND
33 North Dakota									
34 Ohio									NC
35 Oklahoma									
36 Oregon									
37 Pennsylvania			2				3	4	9
38 Rhode Island									
39 South Carolina	1								1
40 Tennessee									NR
41 Texas									NC
42 Vermont									
43 Virginia									
44 Washington									
45 West Virginia									ND
46 Wisconsin									
47 Wyoming									
48 Puerto Rico									
49 Virgin Islands									

Table 13.08: Summation of Confirmed FSD Cases (SD-Disease) by Days from Birth Until Diagnosis

State/Territory	0-15	16-30	31-45	46-60	61-75	76-90	> 90	Unknown	Total
1 Alabama									
2 Arizona									
3 Arkansas									
4 California									
5 Colorado									
6 Connecticut									
7 Delaware									NC
8 District of Columbia									
9 Florida									
10 Georgia									ND
11 Hawaii									
12 Illinois									
13 Indiana									
14 Iowa									
15 Kansas									
16 Kentucky NR									
17 Louisiana									ND
18 Maine									
19 Maryland									
20 Massachusetts									
21 Michigan									
22 Minnesota									
23 Mississippi									
24 Missouri									
25 Montana									NC
26 Nebraska									
27 Nevada									
28 New Hampshire									
29 New Jersey									
30 New Mexico									
31 New York									ND
32 North Carolina									ND
33 North Dakota									
34 Ohio									NC
35 Oklahoma					1				1
36 Oregon									
37 Pennsylvania									
38 Rhode Island									
39 South Carolina									
40 Tennessee									NR
41 Texas									NC
42 Vermont									
43 Virginia									
44 Washington									
45 West Virginia									ND
46 Wisconsin									
47 Wyoming									
48 Puerto Rico									
49 Virgin Islands									

Table 13.09: Summation of Confirmed FAS Cases (Sickle Cell Trait) by Days from Birth Until Diagnosis

State/Territory	0-15	16-30	31-45	46-60	61-75	76-90	> 90	Unknown	Total
1 Alabama									
2 Arizona									
3 Arkansas									
4 California									NC
5 Colorado								278	278
6 Connecticut									
7 Delaware									NC
8 District of Columbia									
9 Florida									ND
10 Georgia									
11 Hawaii								83	83
12 Illinois									
13 Indiana									
14 Iowa									
15 Kansas								126	126
16 Kentucky NR									
17 Louisiana	1,788		39	10	5	15			1,857
18 Maine	10								10
19 Maryland									
20 Massachusetts									
21 Michigan									
22 Minnesota									
23 Mississippi			1	1	1				3
24 Missouri									
25 Montana									NC
26 Nebraska	88	7	1	5	1		2		104
27 Nevada		307							307
28 New Hampshire									
29 New Jersey				2					2
30 New Mexico									
31 New York									NC
32 North Carolina									ND
33 North Dakota									
34 Ohio								636	636
35 Oklahoma									NC
36 Oregon									NA
37 Pennsylvania									
38 Rhode Island									
39 South Carolina									DA
40 Tennessee									NR
41 Texas									NC
42 Vermont									
43 Virginia									
44 Washington									
45 West Virginia		1							1
46 Wisconsin	609								609
47 Wyoming									
48 Puerto Rico	517	500							1,017
49 Virgin Islands	145								145

Table 13.10: Program Follow-up

State/Territory	Program have an Aggressive Follow-up Program for Sickle Carriers	Program has Extra Staff Dedicated to Sickle Carrier Follow-up	Sickle Follow-up Performed for Carriers
1 Alabama	Yes	Yes	Community Based Sickle Cell Organization, Educational Services, Private Medical Providers.
2 Arizona	Yes	Yes	SC disease and carriers referred to AZ Sickle Cell Program for follow-up.
3 Arkansas	Yes	Yes	Parents of carriers offered in home visits and written materials by trained trait counselor.
4 California	No	Yes	Free counseling and testing offered to all families. Additional reminder letters mailed.
5 Colorado	No	No	In home counseling offered and fee for both trait and diagnosis.
6 Connecticut	No	No	Letter sent to primary care provider. A letter and brochure (in Spanish and English) is to the mom.
7 Delaware	No	No	Letters to parent and primary care provider after both 1st and 2nd screens.
8 District of Columbia	Yes	Yes	Follow-up is carried out through DC MCH.
9 Florida	No	No	Trait letters sent to family to encourage further testing & contact with physician.
10 Georgia	No	No	Parents sent letter for counseling by Georgia Sickle Cell Foundation or health dept.
11 Hawaii	No	No	At Tripler Army Medical Center, letter & a sickle cell trait informational booklet are sent to the family whose child is identified to have sickle cell trait.
12 Illinois	No	No	Families of infants w/trait status are referred to a pediatric hematologist and / or public health department to provide education to the families.
13 Indiana	No	No	
14 Iowa	Yes	Yes	
15 Kansas	No	No	
16 Kentucky	No	No	Report only; explain where counseling can be obtained.
17 Louisiana	Yes	No	Letters are sent to parent/guardian w/results indicating sickle carrier status.
18 Maine	No	No	
19 Maryland	No	No	
20 Massachussetts	Yes	Yes	In home counseling and carrier testing offered.
21 Michigan	Yes	Yes	Carrier parents are aggressively followed up, taped counseling sessions, quality assurance.
22 Minnesota	No	No	
23 Mississippi	Yes	Yes	Field staff available for counseling carriers (RN's & Genetic Social Workers).
24 Missouri	Yes	No	Notification letters, follow-up letters and educational genetic counseling.
25 Montana	No	No	None
26 Nebraska	No	No	Follow-up letters sent to parents and physician, along w/pamphlet & fact sheet to parents.
27 Nevada	No	No	Written notification to Primary Care Physician.
28 New Hampshire	No	No	NH Bureau SMS program offers followup testing, counseling & care coord to families on request.
29 New Jersey			
30 New Mexico	Yes	No	Follow-up letters sent to Sickle cell council and parents.
31 New York	No	No	
32 North Carolina	No	No	
33 North Dakota	Yes	No	
34 Ohio			
35 Oklahoma	No	No	All carriers are referred to the Oklahoma Sickle Cell Association.
36 Oregon	No	No	Pamphlet describing traits is provided to the primary care provider to give to the family.
37 Pennsylvania	No	No	Lists of SC traits mailed to treatment centers, center notifies parents, offer counseling.
38 Rhode Island	No	No	
39 South Carolina	No	No	Follow-up by the Sickle Cell community based organization.
40 Tennessee	Yes	No	Counseling of families affected.
41 Texas	No	No	Reports are sent to submitter & submitter is responsible for following carriers.
42 Vermont	No	No	Counseling offered - pediatric hematologist.
43 Virginia	No	No	Parents are sent follow-up letters & toll free number for questions.
44 Washington	No	Yes	A letter & educational materials sent to child's health care prov. when 2 screens are matched. A request for 2nd screen is made to the provider when there is only one screen.
45 West Virginia	No	No	Suggest genetic counseling by provider.
46 Wisconsin	No	No	Family education provided by health educators, pediatric practices, clinics & public health dept. & post natal home visits.
47 Wyoming	No	No	
48 Puerto Rico	Yes	Yes	Letter and appointment for counseling clinic.
49 Virgin Islands	Yes	Yes	

Table 13.11: Summation of Years Covered and Number of Cases Detected by Program

State/Territory	FS: [Probable SCD (May be Hb-S-B-Thalassemia), Hb S-B-thalassemia with hereditary persistent fetal hemoglobin]			FSC: (Presumably Sick-Hemoglobin C Disease)			FSA [Probable Hb-S-B-Thalassemia (Maybe Hb-S with Transfusion)]		
	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases
1 Alabama			NA			NA			NA
2 Arizona	1994	6	29	1994	6	14	1994	6	3
3 Arkansas	1988	12	170	1988	12	96	1988	12	8
4 California	1990	10	782	1990	10	355	1990	10	87
5 Colorado									
6 Connecticut	1989	11	136	1989	11	67	1989	11	8
7 Delaware	1993	7	25	1993	7	17	1993	7	4
8 District of Columbia	1996	4	82	1996	4	40	1996	4	1
9 Florida	1988	12	955	1988	12	525	1988	12	416
10 Georgia NR									
11 Hawaii	May-93	7 1/2	5	May-93	7 1/2	2	May-93	7 1/2	0
12 Illinois	1989	11	1,246	1989	11	477	1989	11	112
13 Indiana	Jul-85	15 1/2	247	Jul-85	15 1/2	118	Jul-85	15 1/2	N/D
14 Iowa NR									
15 Kansas NR				1992	8	2	1999	1	3
16 Kentucky NR									
17 Louisiana	1995	5	285	1995	5	132	N/C		N/C
18 Maine NR									
19 Maryland	1983	17	492	1989	11	264	1989	11	53
20 Massachussetts NR									
21 Michigan	1987	13	575	1987	13	378			
22 Minnesota NR									
23 Mississippi NR									
24 Missouri NR									
25 Montana	2000	0	0	2000	0	0	2000	0	0
26 Nebraska	Nov-96	4 1/6	10	Nov-96	4 1/6	3	Nov-96	4 1/6	0
27 Nevada NR									
28 New Hampshire	1990	10	2	1990	10	1	1990	10	0
29 New Jersey NR									
30 New Mexico	1988	12	0	1988	12	0	1995	5	0
31 New York	1975	25	3,493	1975	25	1,738	1993	7	79
32 North Carolina NR									
33 North Dakota NR									
34 Ohio NR									
35 Oklahoma	1991	9	89	1991	9	44	NC		NC
36 Oregon NR									
37 Pennsylvania NR									
38 Rhode Island NR									
39 South Carolina	1996	4	200	1996	4	97	1996	4	27
40 Tennessee NR									
41 Texas	1983	17	1,312	1983	17	645	1983	17	119
42 Vermont	Mar-90	10 3/4	0	Mar-90	10 3/4	1	Mar-90	10 3/4	0
43 Virginia	1989	11	449	1989	11	252	1989	11	60
44 Washington	May-91	9 1/2	36	May-91	9 1/2	17	May-91	9 1/2	20
45 West Virginia NR				ND		ND	ND		ND
46 Wisconsin	1989	11	148	1989	11	84	1989	11	21
47 Wyoming	Apr-79	20 3/4	2						
48 Puerto Rico	1987	13	NA	1987	13	NA	1987	13	NA
49 Virgin Islands NR									

Table 13.11: Summation of Years Covered and Number of Cases Detected by Program (continued)

State/Territory	FC: [Probable Homozygous Hb-C (May be Hb-C-β-Thalassemia)]			FE: [Probable Homozygous Hb-E (May be Hb-E-β-Thalassemia)]			F Only: (Possible Homozygous β-thalassemia or Normal Biologic Variation)		
	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases
1 Alabama NR									
2 Arizona	1995	5	3	1995	5	3	1995	5	1
3 Arkansas	1988	12	23	1988	12	11	NA		NA
4 California	1990	10	80	1990	10	48 a	1990	10	46
5 Colorado NR									
6 Connecticut	1989	11	20	1989	11	24	1989	11	3
7 Delaware	1993	7	8	1993	7	0	1993	7	10
8 District of Columbia	1996	4	12	1996	4	0	1996	4	11
9 Florida	1988	12	131	ND		ND	ND		ND
10 Georgia NR									
11 Hawaii	May-93	7 1/2	1	May-93	7 1/2	5	May-93	7 1/2	1
12 Illinois	1989	11	70	1989	11	36			
13 Indiana	Jul-85	15 1/2	36	ND		ND	ND		ND
14 Iowa NR									
15 Kansas NR									
16 Kentucky NR									
17 Louisiana	1995	5	44	N/C		N/C	N/C		N/C
18 Maine NR									
19 Maryland							1987	13	6
20 Massachussetts NR									
21 Michigan NR									
22 Minnesota NR									
23 Mississippi NR									
24 Missouri NR									
25 Montana	2000	0	0	2000	0	0	2000	0	0
26 Nebraska	Nov-96	4 1/6	0	Nov-96	4 1/6	1	Nov-96	4 1/6	0
27 Nevada NR									
28 New Hampshire	1990	10	59	1987	13	15	1987	13	4
29 New Jersey NR									
30 New Mexico	1988	12	0	1988	12	0	1988	12	2
31 New York	1975	25	510	1993	7	43	1993	7	5
32 North Carolina NR									
33 North Dakota NR									
34 Ohio NR									
35 Oklahoma	1991	9	13	1991	9	2	NC		NC
36 Oregon NR									
37 Pennsylvania NR									
38 Rhode Island NR									
39 South Carolina	1996	4	15	1996	4	8	1996	4	2
40 Tennessee NR									
41 Texas	1983	17	145	1983	17	160	1983	17	9
42 Vermont	Mar-90	10 3/4	0	Mar-90	10 3/4	0	Mar-90	10 3/4	0
43 Virginia	1989	11	66	1989	11	29	1989	11	15
44 Washington	May-91	9 1/2	7	May-91	9 1/2	142	1991	9 1/2	2
45 West Virginia	ND		ND	ND		ND	ND		ND
46 Wisconsin	1989	11	14	1989	11	35	1989	11	2
47 Wyoming NR									
48 Puerto Rico	1987	13	NA	1987	13	NA	1987	13	NA
49 Virgin Islands NR									

a = E/β-thalassemia only.

Table 13.11: Summation of Years Covered and Number of Cases Detected by Program (continued)

State/Territory	FAS: [Presumably Sickie-Carrier (Trait)]			FAC: [Presumably Hb-C Carrier (Trait)]			FAE: [Presumably Hb-E Carrier (Trait)]		
	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases
1 Alabama						NA	N/A		N/A
2 Arizona	1994	6	1898	1995	5	355	1997	3	62
3 Arkansas	DA			DA			DA		
4 California	N/C		N/C	N/C		N/C	N/C		N/C
5 Colorado NR									
6 Connecticut	1989	11	5,969 a	1989	11	1,729 a	1989	11	192
7 Delaware	1993	7	1,590	1993	7	518	1993	7	18
8 District of Columbia	1996	4	3,373	1996	4	920	1996	4	0
9 Florida	N/D		N/D	N/D		N/D	N/D		N/D
10 Georgia NR									
11 Hawaii	May-93	7 1/2	457	May-93	7 1/2	119	May-93	7 1/2	187
12 Illinois									
13 Indiana	Jul-85	15 1/2	9,454	Jul-85	15 1/2	3,049	DA		DA
14 Iowa NR									
15 Kansas NR									
16 Kentucky NR									
17 Louisiana	N/R		N/R	N/R		N/R	N/A		N/A
18 Maine NR									
19 Maryland NR									
20 Massachusetts NR									
21 Michigan	1987	13	24,706	1987	13	7,352	1987	13	0
22 Minnesota NR									
23 Mississippi NR									
24 Missouri NR									
25 Montana NR									
26 Nebraska	Nov-96	4 1/6	274	Nov-96	4 1/6	80	Nov-96	4 1/6	29
27 Nevada NR									
28 New Hampshire	1990	10	NA	1990	10	NA	1990	10	NA
29 New Jersey NR									
30 New Mexico NR									
31 New York	1975	25	114,738	1975	25	3,954	1987	13	2,034
32 North Carolina NR									
33 North Dakota NR									
34 Ohio NR									
35 Oklahoma	1991	9	3,991	1991	9	1,299	NC		NC
36 Oregon NR									
37 Pennsylvania NR									
38 Rhode Island NR									
39 South Carolina	DA	DA	DA	DA	DA	DA	DA	DA	DA
40 Tennessee NR									
41 Texas	NC		NC	NC		NC	NC		NC
42 Vermont	Mar-90	10 3/4	59	Mar-90	10 3/4	16	Mar-90	10 3/4	11
43 Virginia	1989	11	NA	1989	11	NA	1989	11	NA
44 Washington	May-91	9 1/2	3,311	May-91	9 1/2	830	May-91	9 1/2	1,913
45 West Virginia	ND		ND	ND		ND	ND		ND
46 Wisconsin	1989	11	7,302	1989	11	2,188	1989	11	358
47 Wyoming	Apr-79	20 3/4	9	Apr-79	20 3/4	6	Apr-79	20 3/4	2
48 Puerto Rico	1987	13	NA	1987	13	NA	1987	13	NA
49 Virgin Islands NR									

a = traits are not confirmed by program.

Table 13.11: Summation of Years Covered and Number of Cases Detected by Program (continued)

State/Territory	FAD or FAG: [Presumably Hb-D or Hb-G (Trait)]			FA + Other [Presumably Other Carrier (Probably insignificant clinically but not known w/o further study)]		
	Year Started	Total Years	Total Cases	Year Started	Total Years	Total Cases
1 Alabama NR						
2 Arizona	1996	4	104	1998	2	184
3 Arkansas	DA			DA		
4 California NR						
5 Colorado NR						
6 Connecticut	1998	2	32	N/C		N/C
7 Delaware	1993	7	13	1993	7	13
8 District of Columbia	1996	4	0	1996	4	86
9 Florida	N/D		N/D	N/D		N/D
10 Georgia NR						
11 Hawaii	May-93	7 1/2	9	May-93	7 1/2	1,449 a
12 Illinois NR						
13 Indiana	DA		DA	DA		DA
14 Iowa NR						
15 Kansas NR						
16 Kentucky NR						
17 Louisiana				N/R		N/R
18 Maine NR						
19 Maryland NR						
20 Massachusetts NR						
21 Michigan	1987	13	0	1987	13	0
22 Minnesota NR						
23 Mississippi NR						
24 Missouri NR						
25 Montana NR						
26 Nebraska	Nov-96	4 1/6	15	Nov-96	4 1/6	30
27 Nevada NR						
28 New Hampshire	1990	10	NA	1990	10	NA
29 New Jersey NR						
30 New Mexico NR						
31 New York	1987	13	1,840	1987	13	28,528
32 North Carolina NR						
33 North Dakota NR						
34 Ohio NR						
35 Oklahoma	1991	9	295	NC		NC
36 Oregon NR						
37 Pennsylvania NR						
38 Rhode Island NR						
39 South Carolina	DA	DA	DA	DA	DA	DA
40 Tennessee NR						
41 Texas	NC		NC	NC		NC
42 Vermont	Mar-90	10 3/4	9	Mar-90	10 3/4	62
43 Virginia	1989	11	NA	1989	11	N/A
44 Washington	May-91	9 1/2	252	May-91	9 1/2	2,354
45 West Virginia	ND		ND	ND		ND
46 Wisconsin	1989	11	168	1989	11	973
47 Wyoming	Apr-79	20 3/4	4	Apr-79	20 3/4	14
48 Puerto Rico	1987	13	NA	1987	13	NA
49 Virgin Islands NR						

a = Hgb Bart's.

Appendix

Persons Verifying Data in National Newborn Screening Report - 2000

State/Territory	Data Responders	Laboratory Director Approval	MCH Director Approval
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Newborn Screening Programs in the U.S.

Newborn screening programs exist in all 50 states and the District of Columbia. Each of these 51 programs has identified an individual contact person most knowledgeable about program and followup issues and a second individual whose responsibility is laboratory protocol. There are a number of state programs with either contract laboratories or multiple laboratory sites. In these states, the contact person listed remains the individual deemed most appropriate to address the laboratory issues for the state program. A few states prefer followup issues and laboratory issues to be handled by the same individual and those are so noted.

This listing is intended to be updated as needed and, therefore, individuals detecting errors and requesting changes are asked to write or fax Dr. Brad Therrell, 1912 W. Anderson Lane #210, Austin, TX 78757 (FAX 512-454-6509).

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Newborn Screening Programs in the U.S.

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